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Challenges in healthcare decision-making for rare diseases: exploring systemic heterogeneity and resource allocation in Chile

Nicolás Schöngut-Grollmus[a] ✉ | Fernanda Manríquez-Castillo[b] | Nicolás Olivares-Marchant[a] | Pedro Álvarez-Muñoz[a] | María-Alejandra Energici[c]

[a]Universidad de O'Higgins, Chile.
[b]Universidad Diego Portales, Chile.
[c]Universidad Andrés Bello, Chile.

Abstract Rare diseases, or orphan diseases, affect fewer than 1 in 2,000 people, presenting unique challenges for healthcare systems due to their low prevalence. This study explores the complexities of healthcare decision-making in Chile, focusing on two interrelated themes: the heterogeneity of healthcare systems and the allocation of resources. Using reflexive thematic analysis of interviews with ten healthcare professionals specializing in rare diseases, the study investigates systemic and structural barriers affecting care. Findings reveal that the heterogeneity of Chile's healthcare system creates disparities in diagnosis and treatment, particularly for patients in rural areas where resources are limited. Patients often experience a prolonged "diagnostic odyssey" due to healthcare professionals' lack of knowledge and experience in identifying rare conditions. Resource allocation further complicates care, as decisions are frequently dictated by limited availability rather than clinical best practices. Legislative gaps, such as the restricted scope of the Ricarte Soto Law, exacerbate inequities by limiting access to essential treatments. This paper concludes by recommending targeted policy reforms, enhanced training for healthcare professionals, and the implementation of standardized diagnostic and treatment guidelines. These measures aim to address disparities and improve healthcare outcomes for individuals living with rare diseases, fostering a more equitable healthcare system in Chile.

Keywords: rare diseases, healthcare decision-making, diagnostic odyssey, resource allocation, chilean healthcare system

1. Introduction

Rare diseases, also known as orphan diseases, are health conditions characterized by their low statistical frequency, affecting fewer than 1 in 2,000 people, according to European Union criteria (Encina et al., 2019). Their low prevalence is not merely anecdotal; instead, it reorganizes a series of networks and relationships that differentiate them from more prevalent health conditions (Schöngut-Grollmus & Energici, 2021). This is due to three key elements:

First, there is a tendency for those with rare diseases to have a significantly shorter life expectancy compared to the rest of the population. Between 7,000 and 8,000 rare diseases have been identified, and 80% of these are of genetic origin, with about 50% manifesting during childhood (Repetto Lisboa, 2017). Furthermore, treatment options for these conditions are often limited and focus primarily on palliative care or improving quality of life rather than resolving the underlying health issue, leaving many needs unmet (Hollin et al., 2017). In this sense, it is essential to highlight that non-pharmacological therapeutic alternatives, such as physical therapy, occupational therapy, psychotherapy, and other interventions, can help manage symptoms or improve quality of life (Kumar; Jim, 2010). These should be considered in the medical or health decision-making process, although they do not result in the patient's total recovery.

Second, the doctor-patient relationship, critical to managing health conditions (Charles et al., 1999), is profoundly altered. Classical literature on the doctor-patient relationship recognizes four models (Emanuel & Emanuel, 1992):

- The paternalistic model is where the doctor uses their expertise to make decisions about the patient, treatments, and interventions.
- The informative model is where the doctor neutrally presents different treatment options, outlining the pros and cons, and the patient makes the final decision.
- The interpretative model is where the doctor seeks to understand the patient's desires and intentions to offer medical interventions tailored to them.
- The deliberative model is where the doctor helps the patient choose the best option based on their clinical situation.

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However, the doctor-patient relationship in rare diseases does not fit neatly into these models (Budych et al., 2012). These models assume that the doctor has enough experience and expertise to diagnose, treat, and manage the patient's condition, which is not always true with rare diseases. Often, doctors lack experience in this category of conditions, and patients, their caregivers, and families, due to their cultural and social capital, may know more about the condition. Moreover, patients and families gain experience from navigating the healthcare system chronically. As a result, in rare diseases, it is common for patients, families, and caregivers to have more knowledge and experience about the condition than the doctor, altering the typical doctor-patient dynamic. The lack of familiar interactions between doctors and patients or a straightforward way to make medical decisions in the case of rare diseases increases feelings of uncertainty, ambiguity, and unpredictability regarding the decision-making process (Budych et al., 2012).

Thirdly, the issue of small markets and low statistical power in the context of rare diseases must be considered, as it affects the development of new therapeutic options for these conditions. Unlike more common diseases, which have large economic markets, the pharmaceutical industry invests in treatments and medications for these ailments due to the high expected financial return. Additionally, common diseases allow for clinical trials with a large number of participants, facilitating the production of evidence on the effectiveness of treatments and increasing the likelihood of approval by regulatory agencies such as the FDA (Food and Drug Administration) in the United States and the EMA (European Medicines Agency) (Nony et al., 2014). This also supports the development of clinical guidelines for evidence-based medicine.

In contrast, the situation is different with rare diseases due to their low prevalence. Markets for these pharmacotherapies are small, and clinical trials, with small sample sizes, lack the necessary statistical power, making approval more difficult (Nony et al., 2014). As a result, research and the development of therapies for rare diseases are often driven by patient organizations rather than the pharmaceutical industry (Bhattacharya et al., 2019). This has led to less scientific evidence on medications, limiting the available treatment options and the development of clinical guidelines for adequately managing these conditions.

The uncertainty caused by the disruption of these three conditions is not only experienced by rare disease patients, their families, and caregivers (Schöngut-Grollmus et al., 2021) but also by the healthcare and non-healthcare professionals working with these communities. It is not only the patients who suffer from the lack of knowledge and recognition of rare diseases, but it also involves a broader transformation of the entire healthcare network concerning rare diseases as a phenomenon and category (Schöngut-Grollmus & Energici, 2021). In this sense, rare or orphan diseases become an essential object of study to observe the transformations experienced by different nodes within healthcare systems.

At the same time, international literature propose as well that rare diseases present unique challenges in healthcare systems, affecting diagnosis, treatment, and patient care (Hay et al., 2020; Tumienė & Graessner, 2021). These conditions often lead to long diagnostic odysseys, limited access to treatments, and complex care coordination burdens for patients and families (Tumienė & Graessner, 2021). Social media research in rare diseases, while promising, faces limitations in participant representativeness (Miller et al., 2021). The development of alternative pharmaceutical innovation pathways, termed social pharmaceutical innovation, is proposed to address unmet needs (Douglas et al., 2022). Patients with rare diseases experience significant psychosocial and economic burdens, emphasizing the need for emotional and companionship support (Smits et al., 2022). Despite progress in diagnostic capabilities and therapy development, 90% of rare diseases lack effective treatments (Groft et al., 2021). Comprehensive studies on the impact of rare diseases across countries are needed, as existing research often lacks a holistic approach (Delaye et al., 2022).

Given the above, this article focuses on how professionals involved in health decision-making handle rare diseases and why managing this phenomenon is crucial. How healthcare professionals manage rare diseases is vital because it directly affects the quality of care patients receive and the healthcare system's ability to adapt to the unique challenges posed by these conditions.

2. Materials and Methods

This article derives from a more extensive project to understand how health decisions are made regarding rare diseases in the Chilean context. The project consisted of three studies focusing first on health decision-making in health professionals who work with rare diseases, a second study regarding the same topic but with patients and carers, and a third study concerned with hybrid forums between patients, professionals, policymakers, and pharmaceutical industry representatives and extended between 2022 and 2024. The project was designed through a qualitative approach to produce knowledge through the conceptualization of healthcare decisions in the context of rare and orphan diseases (Krause, 1995) instead of their quantification. As Krause states (1995, p. 21), qualitative research refers to procedures that allow the construction of knowledge that uses concepts as a basis. Concepts enable the reduction of data complexity in qualitative research. Using concepts, we can establish the coherence and comprehension that a given scientific product needs to be communicated and understood in social sciences. This paper tackles the first study's results about health decision-making in professionals who work with rare diseases.

For this purpose, we recruited ten participants described in Table 1.

Table 1 Characterization of participants of this study.

Identifier	Profession	Relation to rare diseases
Participant 1	Geneticist	Clinician at private and public hospitals and researcher
Participant 2	Infectious disease doctor	Clinician at public hospital and researcher
Participant 3	Genetic counselor	Clinician at a private hospital
Participant 4	Physiatrist	Clinician at public hospital
Participant 5	Nurse	Lecturer and employee at a startup related to rare diseases
Participant 6	Clinical psychologist	Therapist at public hospital
Participant 7	Traumatologist	Clinician at public hospital
Participant 8	Occupational therapist	Private practice
Participant 9	Medical Technologist	Manager of rare disease office at public hospital
Participant 10	Lawyer	Specialized in health law and rare diseases

Source: Own source.

We employed semistructured interviews as our data collection method, as this approach has effectively examined participant's views and decision-making processes concerning medical and healthcare matters (Allen et al., 2019).

We used Reflexive Thematic Analysis (Braun & Clarke, 2006, 2014), an inductive technique for identifying recurring patterns and themes within the qualitative data collected from the interviews. All interviews were conducted either via video conferencing platforms or in-person. All of them were audio recorded, and subsequently transcribed to facilitate thorough analysis. The analytical process adhered to Braun and Clarke's (2006) guidelines. Initially, we immersed ourselves in the interview data to familiarize ourselves. Next, we performed open coding, utilizing both in vivo and descriptive codes to highlight elements that could help answer our research questions and meet our objectives. Following this, the research team engaged in internal discussions to compare and refine the codes, which led to the development of preliminary themes. These were then cross-checked against the data, identifying two primary themes discussed in the Results section. In the fourth step, we defined and named these themes before crafting their narrative write-up in the final stage. This write-up aims to convincingly demonstrate the rigor and credibility of our analysis (Braun and Clarke, 2006) and to present an engaging analytical story that captures the essence of our data (Braun and Clarke, 2006).

3. Results

The thematic analysis of the ten interview transcripts led us to propose two themes that group the content throughout the interviews: the heterogeneity of healthcare systems for rare diseases in Chile and the allocation of resources as the primary criterion for health decision-making in rare diseases.

3.1. Heterogeneity of healthcare systems for rare diseases

The care of rare diseases faces an inherent challenge due to the great diversity of these pathologies and their low prevalence. A rare disease is estimated to affect less than one person per 2,000 inhabitants. In a population like Chile's, people with rare diseases represent a tiny fraction of the patients cared for by the health system, even though as a category, they make up to approximetly 6% of the Chilean population (Repetto Lisboa, 2017). This low prevalence brings a series of structural, logistical, and medical complications that exacerbate the difficulties faced by patients, their families, and the health professionals responsible for their treatment. In this context, a significant heterogeneity in healthcare systems for rare diseases is manifested, which can be broken down into four aspects:

3.1.1. Disease trajectory

One of the most critical problems patients face with rare diseases is the diagnostic process, known as the "diagnostic odyssey." This "odyssey" refers to the series of consultations, exams, tests, and referrals that affected individuals must undergo before receiving a definitive diagnosis. Since rare diseases often present uncommon or atypical symptoms, general practitioners or even specialists in common areas of medicine may struggle to recognize the signs of these pathologies. This generates a prolonged process where patients may spend years searching for answers about their health condition. In some cases, the search for a diagnosis can extend up to five years, which not only prolongs the physical and psychological suffering of patients but also places a heavy burden on families and caregivers, who face high levels of uncertainty and stress.

The diagnostic process is not homogeneous. It largely depends on the doctor's or healthcare professional's knowledge of the patient and the resources available at the institution where the care is provided. Patients often go through multiple specialists before obtaining a diagnosis, and these professionals often struggle to fit the symptoms into a specific pathology due to a lack of information and training on rare diseases. It is essential to highlight that this is not a lack of training at the individual level of the doctor but rather a broader structural problem due to the rarity of these pathologies and the large number of different diseases that fall under the category of rare diseases, it is practically impossible for a general practitioner or even a specialist to have in-depth knowledge of all of them.

3.1.2. Inequality in the availability of treatments and legislation

In addition to the difficulty of diagnosis, rare diseases also present significant variability in the treatments available and access to them. This variability is especially evident in the financing systems and legislation covering these diseases' treatments.

In Chile, for example, only some rare diseases are covered by the Ricarte Soto Law, a law that funds high-cost disease treatments, but this coverage is limited and includes only a few rare pathologies. This means that many patients with rare diseases must face the reality that the treatment they need is either unavailable or highly costly. When they exist, treatments tend to be expensive. This creates a situation of inequity, as people with common diseases can access more affordable treatments while those with rare diseases face insurmountable economic barriers.

3.1.3. Technical and professional training

Another critical aspect of the heterogeneity in healthcare systems for rare diseases is the variability in the technical and professional training available at different healthcare centers. In many cases, patients with rare diseases face a lack of trained professionals who can recognize and treat their pathologies. This lack of training is partly due to the low prevalence of rare diseases and specific training in this area within medical school curricula. Many healthcare professionals do not have the necessary training to identify the signs of a rare disease and, therefore, may not refer the patient to an appropriate specialist or perform the necessary tests to reach a diagnosis.

The variability in the availability of training and education affects doctors and other healthcare professionals who play a fundamental role in treating rare diseases, such as nurses, psychologists, social workers, and physiotherapists. The comprehensive care of a person with a rare disease often requires the intervention of a multidisciplinary team, and the lack of training of these professionals exacerbates the difficulties already faced by patients and their families.

3.1.4. Disparity in technical and technological resources

Heterogeneity is also manifested in technical and technological resources available at different healthcare centers. Not all hospitals and clinics in Chile have the equipment to perform the advanced diagnostic tests required for some rare diseases. This forces patients to travel to larger centers or wait months to undergo tests crucial to obtaining a definitive diagnosis.

The lack of homogeneity in the availability of resources is not limited to technical equipment but also extends to the availability of specific medications and treatments. Some healthcare centers have access to advanced treatments, while others cannot offer these therapies, creating inequities in healthcare access.

3.2. Allocation of resources in rare diseases

Another key focus of this analysis is how financial and human resources are distributed within the healthcare system and how this distribution affects the treatment of rare diseases. The allocation of resources is one of the most critical factors determining the quality of care and the decisions made in rare disease cases.

3.2.1. Unequal geographical distribution

One of the most critical factors in resource allocation is geographical disparity. Chile has an extensive and varied geography, directly affecting resource availability in different regions. While in large cities like Santiago, patients may have access to specialized centers for rare diseases, people living in rural or remote areas face significant barriers to accessing these services. This includes the lack of specialists in rural areas and the absence of adequate infrastructure to perform diagnostic tests or carry out complex treatments.

Patients from rural areas often have to travel long distances to receive specialized care, which entails additional transportation, accommodation, and time costs. Additionally, doctors in these areas often lack the training and resources to diagnose and treat rare diseases, further prolonging the "diagnostic odyssey."

3.2.2. Economic limitations

The cost of treatments for rare diseases is a critical factor affecting the decision-making process for patients and healthcare professionals. As mentioned earlier, treatments for rare diseases are often costly, and health systems often do not cover these costs in many cases. Patients and their families are forced to resort to fundraising campaigns, loans, or even seek treatment in other countries, further aggravating the emotional and financial burden they already face.

The allocation of financial resources also affects the availability of treatments at the institutional level. Hospitals and clinics that lack the necessary resources to acquire the medications or carry out the treatments required by patients with rare diseases must make difficult decisions about allocating the available funds. This may mean that patients with rare diseases are relegated to the background in favor of other patients with more common diseases or those requiring less expensive treatments.

3.2.3. Decision-making based on resource availability

A central aspect of resource allocation is that many medical decisions in rare disease cases are made based on the availability of these resources. That is, the treatment offered to a patient is not always based on what would be clinically ideal but rather on what is possible within the limitations of the healthcare system.

For example, a patient living in a region without access to a rare disease specialist may receive suboptimal treatment simply because no options are available. Similarly, a patient whose treatment requires a medication that is not included in the public financing system may have to resort to alternative options that may not be as effective or may not be fully available.

Healthcare professionals also face challenges when making decisions in these contexts. The lack of adequate resources, both in terms of training and infrastructure, means that they often have to improvise solutions to address their patients' health problems. This can result in subjective decision-making based on the physician's personal experience rather than on established protocols or scientific evidence.

4. Discussion

This study explores the complexities of health decision-making processes for rare diseases in Chile, mainly focusing on the heterogeneity of healthcare systems and the allocation of resources as key factors influencing outcomes for patients with rare conditions. The results highlight essential challenges and disparities that must be addressed to improve healthcare provision for individuals with rare diseases.

As highlighted in this study, the heterogeneity of healthcare systems for rare diseases reflects the complexity and diversity inherent to these conditions. Rare diseases encompass a wide range of pathologies, most genetic, with many manifesting in childhood. Given the low prevalence of these conditions, healthcare systems often struggle to provide timely and accurate diagnoses, creating what has been referred to as the "diagnostic odyssey". This is a prolonged journey that patients and their families must endure while seeking answers about their health condition. The impact of this diagnostic uncertainty on patients and their families cannot be overstated, as it exacerbates the psychological and emotional burden of living with a rare disease. This is not exclusive to the Chilean reality, as it is an issue in many different countries; for example, literature on this topic points out that many patients with rare diseases experience significant delays in receiving an accurate diagnosis. In the UK, 46% of rare disease patients waited over one year for a definitive diagnosis, with 20% waiting at least five years. In Australia, 51.2% of patients waited one or more years for a diagnosis, with 30% waiting five or more years (Gong et al., 2020).

The diagnostic odyssey affects patients and places significant strain on healthcare professionals. General practitioners and specialists frequently lack the knowledge or resources to effectively diagnose and treat rare diseases. The lack of familiarity with these conditions means that the traditional doctor-patient relationship, typically built on a foundation of medical expertise and trust, is often disrupted. In many cases, patients and their families become more knowledgeable about the disease than the healthcare professionals as they accumulate years of experience navigating the healthcare system. This role reversal can lead to frustration and uncertainty for both patients and professionals, further complicating the decision-making process. Again, this situation is not solely in the Chilean context. However, other international studies continue to have similar results: Studies by Gong et al. (2020) and Walkowiak and Domaradzki (2021) show that healthcare professionals, especially general practitioners, often lack awareness and knowledge about rare diseases. A study in Poland found that 95.4% of medical students perceived their knowledge about rare diseases as insufficient or very poor (Walkowiak & Domaradzki, 2021). In China, only 5.3% of physicians were moderately or well aware of rare diseases (Gong et al., 2020).

Moreover, the diversity of rare diseases makes it difficult for healthcare systems to establish standardized treatment protocols. The lack of clear guidelines for treating these conditions means that decisions are often made on a case-by-case basis, contributing to unpredictability and inconsistency in care. This finding aligns with previous research emphasizing the need for more comprehensive training and education for healthcare professionals on rare diseases (Budych et al., 2012). Without adequate knowledge, healthcare professionals may be ill-equipped to recognize symptoms, leading to delays in diagnosis and treatment. Similar international studies showed that many health professionals state that they feel ill-prepared to tackle rare diseases (Li et al., 2021; McMullan et al., 2021), and in some cases, this is solved using the internet as the primary source of information for doctors regarding rare diseases (Avellaneda Fernández et al., 2012).

This heterogeneity extends beyond diagnostic and treatment challenges. The study also revealed significant inequalities in the availability of treatments and legislation for rare diseases. In Chile, the Ricarte Soto Law provides some financial support for high-cost treatments, but its coverage is limited, and most rare diseases are not included. This legislative gap leaves many patients without access to necessary treatments, further exacerbating the inequities in healthcare for individuals with rare conditions. While this situation is not unique to Chile, it highlights the broader global challenge of ensuring equitable access to care for individuals with rare diseases. The lack of financial support for treatments also underscores the need for advocacy and policy reform to address the specific needs of this patient population.

The second central theme of the study relates to the unequal distribution of resources for rare diseases, which significantly impacts the quality of care that patients receive. Resource allocation is a critical determinant of health outcomes,

particularly in the case of rare diseases, where the scarcity of resources—both financial and human—can be a significant barrier to effective care.

One of the most critical issues identified in this study is the geographical disparity in resource allocation. Chile's vast and varied geography presents a unique challenge, as patients living in rural or remote areas often face significant obstacles in accessing specialized care. The lack of specialists and infrastructure in these regions means patients must travel long distances for necessary care. This places a financial burden on patients and their families and prolongs the diagnostic odyssey, as access to specialized diagnostic tests and treatments is often delayed.

The unequal distribution of financial resources compounds geographical disparities. The high cost of treatments for rare diseases is a well-documented issue, and this study confirms that many patients in Chile cannot afford the treatments they need. The limited coverage provided by the Ricarte Soto Law means that many families must resort to fundraising campaigns, loans, or even seeking treatment abroad. This creates a situation where the ability to access care is determined by one's financial means rather than medical necessity, contributing to significant inequities in health outcomes.

Moreover, healthcare professionals themselves are often constrained by the availability of resources. The study found that medical decisions for patients with rare diseases are frequently based on the availability of resources rather than what is clinically ideal. This highlights the tension between medical knowledge and resource limitations, where healthcare professionals may be forced to make suboptimal decisions due to a lack of appropriate treatments or equipment. In such cases, decision-making is often subjective, based on the personal experience of the healthcare provider rather than established guidelines or evidence-based practice. This further contributes to the variability and inconsistency in the care of patients with rare diseases.

Another significant finding from the study is the lack of adequate training for healthcare professionals working with rare diseases. Many professionals, particularly those in rural areas, lack the specialized knowledge to diagnose and treat rare conditions. This lack of training prolongs the diagnostic process and affects the quality of care provided. The need for more comprehensive education on rare diseases is evident, as it would help healthcare professionals make more informed decisions and provide better care for their patients.

The findings of this study have important implications for policy and practice. First, there is an urgent need for policy reform to address the gaps in healthcare coverage for rare diseases. The limited coverage provided by existing legislation, such as the Ricarte Soto Law, leaves many patients without access to the necessary treatments. Expanding this coverage to include a broader range of rare diseases and treatments would help alleviate the financial burden on patients and ensure more equitable access to care. This is coherent with other literature on the topic, where while doing fieldwork with patients, caregivers, and healthcare professionals in rare diseases, all stakeholders unanimously agree that healthcare systems are often inadequate in addressing the needs of rare disease patients (Lopes et al., 2018). Regarding health professionals exclusively, they emphasize the need for improved care coordination, patient empowerment, and earlier interventions (McMullan et al., 2021).

Second, there is a need for greater investment in training and education for healthcare professionals. Providing specialized training on rare diseases, particularly for those working in rural or remote areas, would help improve the quality of care for patients and reduce the time it takes to reach a diagnosis. This training should be accompanied by the development of standardized guidelines for diagnosing and treating rare diseases, which would provide healthcare professionals with the tools they need to make more informed decisions.

Finally, addressing the geographical disparities in healthcare access requires a coordinated effort to improve infrastructure and resources in underserved areas. This could involve the development of telemedicine programs to connect patients with specialists in urban centers or establishing regional centers of excellence for rare diseases.

5. Conclusions

This study provides critical insights into the challenges and disparities in healthcare systems for rare diseases, particularly in the Chilean context. The findings highlight two significant themes that significantly influence healthcare outcomes for rare disease patients: the heterogeneity of healthcare systems and the unequal allocation of resources. Both elements contribute to the prolonged "diagnostic odyssey" faced by patients and their families, exacerbating the emotional and financial burdens associated with rare diseases.

The heterogeneity of healthcare systems for rare diseases reflects the diversity of these conditions and the lack of standardized diagnostic and treatment protocols. With rare diseases affecting a small percentage of the population, healthcare professionals often lack the necessary knowledge and experience to provide accurate diagnoses and effective care. This results in a role reversal where patients and families sometimes know more about their condition than the healthcare providers, disrupting the traditional doctor-patient relationship. This is not only a Chilean issue but a global one, as studies from various countries show similar delays in diagnosis and a lack of preparedness among healthcare professionals when dealing with rare diseases.

The study also highlights significant inequalities in the availability of treatments and legislation for rare diseases. In Chile, while some financial support exists through the Ricarte Soto Law, it is limited and does not cover all rare conditions. This

legislative gap leaves many patients without access to necessary treatments, underscoring the need for policy reform to expand coverage and ensure more equitable access to care. Furthermore, the high costs of treatments for rare diseases pose significant financial challenges, pushing many patients to seek alternative solutions such as fundraising or treatment abroad.

The second central theme, the unequal allocation of resources, reveals critical geographical and economic disparities in access to specialized care. Patients in rural or remote areas face significant obstacles in accessing the necessary infrastructure and specialists for proper diagnosis and treatment, further prolonging the diagnostic process. This uneven distribution of resources also affects healthcare professionals, who are often constrained by the availability of treatments and equipment, leading to suboptimal medical decisions. Resource limitations force healthcare professionals to base their decisions not on clinical ideals but on what is practically available, contributing to inconsistency in care.

Based on these findings, the study suggests several critical implications for policy and practice. First, expanding the coverage of rare diseases in health legislation like the Ricarte Soto Law is crucial to alleviate the financial burden on patients and ensure equitable access to treatments. Second, there is an urgent need for more significant investment in the education and training of healthcare professionals, particularly in rural areas, to improve their ability to diagnose and treat rare diseases. Standardized guidelines for rare disease management would provide much-needed support to healthcare providers. Finally, addressing geographical disparities in healthcare access requires coordinated efforts to enhance infrastructure in underserved areas, potentially through telemedicine or establishing regional centers of excellence.

This paper has a few limitations that should be acknowledged to contextualize its findings. First there is a limited number of participants of ten healthcare professionals. This might affect the generalizability of the results, as their experiences may not fully represent the broader spectrum of healthcare providers involved in rare disease management. Additionally, while the inclusion of participants from diverse professional backgrounds enriches the analysis, it may also introduce variability that complicates the synthesis of uniform themes. The use of reflexive thematic analysis, though a robust qualitative method, relies on the researchers' interpretation, which may be influenced by subjective biases or preconceptions. Moreover, the study focuses exclusively on the Chilean healthcare system, which limits the applicability of its findings to other contexts with differing healthcare structures. Finally, this paper does not consider perspectives from patients and caregivers, which is relevant as their lived experiences could provide crucial insights into the "diagnostic odyssey" and the systemic barriers they face. Addressing these limitations in future research—through larger, more diverse samples and the inclusion of multiple stakeholder perspectives—would contribute to a more comprehensive understanding of the challenges in rare disease healthcare decision-making.

In conclusion, this study underscores the need for comprehensive reforms in healthcare systems to address the unique challenges of rare diseases better. By improving resource allocation, expanding policy coverage, and enhancing professional training, healthcare systems can provide more consistent and equitable care, ultimately improving the quality of life for individuals with rare diseases.

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Ethical considerations

This study involves human subjects and adheres to the necessary ethical guidelines for conducting research with such participants. Prior to initiating the study, approval was obtained from two ethics committee, from Universidad Alberto Hurtado first, and from the ethics commitee of the Fundación Arturo López Pérez second, to ensure that all ethical standards were met. Informed consent was obtained from all participants, who were first thoroughly briefed on the nature of the research, its objectives, and potential risks. Participants were provided with clear and comprehensive information, both verbally and in writing, before consenting to take part in the study. Each participant signed a written consent form, confirming their voluntary participation and understanding of the research process. All data collection procedures were conducted in compliance with the ethics commitee protocol, ensuring the confidentiality and well-being of all participants.

Conflict of Interest

The authors declare no conflicts of interest.

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