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The Narrative of Silence: Challenges Faced by Informal Caregivers of Patients with Rare Diseases in China

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Abstract. Rare diseases pose a significant challenge to global public health. Patients with rare diseases and their informal caregivers confront a narrative dilemma, impacting both their well-being and the functioning of the care ecosystem. This study focuses on the phenomenon of the "narrative of silence" among informal caregivers of patients with rare diseases in China. Based on the integration of existing literature and case interviews, this study reveals its specific manifestations and causes. This research finds that the informal caregivers' "narrative of silence" mainly includes three types: "performative silence", "semantic silence", and "power silence", all of which negatively impact their mental health, medical coordination and family relationships. Furthermore, this silence is shaped by the interplay of four factors: the characteristics of rare diseases directly constrain informal caregivers' narrative capacity, mainstream illness narrative preferences and Chinese family characteristics jointly suppress narrative willingness internally, and social proverbs externally restrict narrative opportunities, collectively trapping informal caregivers in a cycle of silence. To address this, recommendations are made in healthcare responsiveness, narrative diversity, support networks, and cultural critique. These measures aim to break the silence by addressing its root causes and fostering a more supportive environment for rare diseases families.

Keywords: Informal Caregivers, Rare Diseases, Narrative of Silence

1. Introduction

Rare diseases have become a significant issue affecting global public health and human well-being. The global population of patients with rare diseases ranges from 263 million to 446 million, but due to statistical differences across regions, rare diseases have long been treated as a low-probability event [1]. In China, over 16.8 million patients with rare diseases suffer from dual neglect: the lack of an official definition and an average diagnostic delay of 4 to 5 years [2,3]. More severely, the informal caregivers, who undertake core and extensive care tasks for patients with rare diseases, have long been voiceless.

Existing research often focuses on patients with rare diseases while neglecting caregivers, and frequently analyzes the narrative of silence phenomenon among patients but overlooks the narrative of silence among informal caregivers. When specifically studying informal caregivers of patients with rare diseases, sample bias often occurs, focusing on online mutual aid groups with higher

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narrative willingness, while ignoring isolated informal caregivers with negative narrative willingness. This leads to an optimistic estimation of the group's narrative willingness and fails to address the phenomenon of "the narrative of silence". When explaining the silence phenomenon in the illness narratives of the Chinese informal caregivers of patients with rare diseases, existing Western illness narrative theories cannot fully explain it. These theories require integration with China's cultural characteristics. Therefore, this paper focuses on the group of informal caregivers of patients with rare diseases in China, aiming to analyze why this group exhibits silence in illness narratives. First, this study will examine the key characteristics, and then explore the factors that lead to such silence. Simultaneously, this study interviews informal caregivers within a Chinese rare disease family to further demonstrate and expand relevant conclusions.

On a theoretical level, this study can compensate for the limits of Western theories by incorporating the characteristics of Chinese family and culture into the illness narrative analysis framework, thereby enriching the localized theoretical framework of narrative medicine. Furthermore, this study focuses on the narrative challenges faced by informal caregivers, aiming to expand the subjects of illness narrative research and build upon existing research conclusions. On a practical level, this study increases attention to the group of informal caregivers of patients with rare diseases, providing a theoretical basis for the formulation of relevant strategies and the improvement of policies. It also offers several recommendations that help enhance informal caregivers' narrative abilities and mental health, while contributing to the development of a more inclusive rare disease care ecosystem.

2. The narratives of informal caregivers of patients with rare diseases

Informal caregivers of patients with rare diseases in China often exhibit a phenomenon of the narrative of silence. When they are asked to discuss or explain the patient's illness experience, informal caregivers tend to remain silent, simplify their expression, avoid in-depth narration, or conceal specific facts. Narrative of silence not only obscures the informal caregivers' actual caring experiences and emotional struggles, exacerbating their social isolation and psychological pressure, but also reduces the quality of medical care for the patients and may negatively impact family and social relationships.

2.1. Informal caregivers of patients with rare diseases

Informal caregivers of patients with rare diseases are typically defined as family members who provide unpaid, continuous assistance with activities of daily living (ADLs) for the patients [4]. They are also the core bearers of the rare disease support system. In rare disease families, informal caregivers mainly consist of adult children, the patient's spouse, and the patient's parents.

They bear significant caregiving responsibilities, covering three major areas: physiological care, medical coordination, and emotional support. They are basic caregivers, providing 24/7 uninterrupted physiological care, including bathing, feeding, and cleaning. They also act as medical coordinators, helping patients navigate the complex diagnosis and treatment systems, despite lacking professional knowledge [5]. After diagnosis, they take on the primary caregiving role once again. Unlike other diseases, rare diseases tend to lack enough support from medical or social systems, and thus rely heavily on family support. That is to say, while patients with rare diseases may receive professional care, they depend more on informal care at home, further increasing the caregivers' pressure.

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Although informal caregivers are required to constantly explain the patient's condition to others and manage their emotions, they often exhibit low willingness and ability to narrate the illness. On one hand, they want to fulfill a role that meets societal expectations, but they fail to align with these expectations through their illness narratives. On the other hand, due to the complexity and general unfamiliarity of rare diseases, informal caregivers find it challenging to provide a precise, specific, and widely accepted account.

In fact, informal caregivers bear the caregiving costs shifted by the healthcare system. They often take on caregiving tasks without support from professional health institutions. They assist with daily care, coordinate medical appointments, and provide emotional support. Paradoxically, when the care recipient is in a nursing home or hospitalized, paid health workers perform these tasks. In contrast, informal caregivers provide these essential services unpaid for patients with rare diseases, positioned outside professional thresholds but forced to fill gaps in the medical system. Their labor is often simplified as a family responsibility rather than being recognized as a part of the formal workforce that deserves institutional acknowledgment.

To illustrate this argument more concretely, this study focuses on a typical rare disease patient family. The patient, a 48-year-old male, suffers from spasmodic torticollis, causing persistent head turning and involuntary neck spasms. This highly evident condition often attracts public attention and misunderstanding. The core informal caregivers are his 47-year-old wife and 21-year-old son, who undertake the vast majority of daily care and emotional support tasks. Furthermore, extended family members such as the patient's parents, mother-in-law, and sisters also participate in care to some extent and influence family decision-making processes, together forming a care network with inter-generational interactions and cultural tensions. The wife and son in this case exhibit all the aforementioned characteristics of informal caregivers, including high-intensity caregiving workload, tendencies to avoid narrating experiences, and a marginal position within the medical system, providing rich observational details for this study to explore the causes.

2.2. The narrative dilemma

To understand the narrative dilemma of informal caregivers, it is first necessary to clarify the concept of "illness narrative": it is a comprehensive description of the illness by individuals or informal caregivers, detailing the process of its occurrence and development, the specifics of care practices, and physical, mental, and emotional experiences. Its essence is to transform fragmented experiences related to the illness into a transmissible and comprehensible system of meaning. For informal caregivers of patients with rare diseases, an ideal illness narrative should fulfill the roles of information transmission, emotional catharsis, and relationship building, serving as a crucial link to establish positive interactions with the outside world and within the family.

However, the illness narratives of informal caregivers of patients with rare diseases present a systematic state of silence. This is manifested not only by a significant reduction in narrative willingness and ability in medical consultations and social relationships, but also by the difficulty their true information, experiences, and emotions face in gaining full expression and recognition within mainstream narrative spaces. The narrative of silence is not simply aphasia or avoidance. It is a kind of narrative alienation caused by the combined effect of structural constraints and individual emotions. Informal caregivers, often due to physical and mental fatigue, ineffective external feedback, and repeated misunderstandings, gradually lose the motivation for systematic articulation and emotional release. The essence of this narrative of silence is a closed loop formed and reinforced by the interaction between limited individual narrative capacity and structurally hindered narration [6]. On the one hand, due to a lack of professional terminology and public channels for

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expression, informal caregivers' narrative abilities are weakened and their opportunities for expression are limited. On the other hand, established societal templates for illness narratives significantly constrain the form and content of their expressions. When informal caregivers express negative emotions that are inconsistent with societal expectations, these expressions are often viewed as incorrect or dismissible.

At the same time, the narratives of silence, in turn, have a series of negative effects on informal caregivers themselves and the families of patients with rare diseases. For informal caregivers, the narratives of silence exacerbate their loneliness and psychological burden. Long-term emotional repression can easily lead to mental health problems such as anxiety and depression, weakening their self-identity and social connections. For the family system, narratives of silence hinder emotional sharing and stress relief among members. This may not only aggravate communication barriers within the family, but also make it difficult for the family as a whole to seek resources and support from the outside, thus falling into deeper isolation and vulnerability. In short, the narrative challenge faced by informal caregivers of patients with rare diseases is not just a problem of personal expression, but a complex problem shaped by multiple factors such as the medical discourse system, the family discourse system, and the social and cultural structure.

2.2.1. Types of the narrative of silence

The narrative of silence can be divided into three types based on the characteristics of their performance, and these three types may occur simultaneously in the same informal caregiver.

The first type is performative silence, which refers to narrative performances that occur to conceal the name of the disease. Informal caregivers remain silent about the reality of the rare disease and choose socially acceptable terms to reinterpret the disease. Silence is therefore both a personal performance and a resistance strategy for rare disease families to confront the disease and its stigma [7]. These performative narratives reveal a paradox: the narrative of silence among informal caregivers of Chinese patients with rare diseases does not imply the complete suppression of illness narration; rather, it reflects an attempt to mitigate family stigma by disguising the disease. Informal caregivers need to remain silent about the family member's health, undertaking the emotional labor of disguising wellness. In the case study, the patient's wife, as the primary informal caregiver, explained the patient's public symptoms as the worsening of a common illness, using the common disease name "cervical spondylosis" to explain to passersby. And when a nurse assisted with botulinum toxin injections, she again disguised the rare disease as "nerve compression," attempting to persuade the doubtful nurse.

The second type, semantic silence, involves avoiding disease-related vocabulary. During the care process of informal caregivers, words referring to the disease name are deliberately replaced. Similar to cancer caregivers' narratives, the narratives of informal caregivers of patients with rare diseases are also influenced by Confucian culture. In the Chinese cultural context, rare diseases are frequently associated with notions of death and fear, which renders their open discussion socially taboo. Consequently, a specific rare disease is often metaphorically referred to as this disease or that disease [8]. In this way, disease names are actively replaced with culturally safe terms, forming a semantic isolation system within the family. Such substitution functions not merely as a linguistic strategy; instead, when the direct articulation of the disease name is perceived as a threat to family harmony, metaphor and concealment of the illness operate as discursive devices to preserve apparent harmony [9]. For instance, in the studied family, the patient's pathological neck tilt made the word "askew" not only denote physical abnormality but also metaphorically imply disorder in the family. Family caregivers avoided words like "askew" and "collapse," treating them as taboos in daily life.

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The third type, power silence, arises when informal caregivers are deprived of the right to speak about the disease by power structures. The imbalance of power between doctors and patients, as well as intergenerational authority within families, jointly suppresses the voice of informal caregivers. On the medical side, patients and caregivers often struggle to participate in decision-making [10]. Informal caregivers, lacking professional medical terminology, find their observations dismissed as "unscientific" by doctors. In the studied rare disease family, the patient's wife employed lay terminology to describe his muscle stiffness during a consultation, yet the clinician dismissed her account as irrelevant, resulting in an additional misdiagnosis. During the referral process, informal caregivers also faced heavy narrative burdens due to frequent misdiagnoses and repeated explanations. The wife, having to explain or conceal the rare disease repeatedly in medical and social contexts, gradually lost personal emotion in the mechanically retold "standardized script." Exhaustion led her to narrative withdrawal, even refusing to recount the illness experience further. Within the family, intergenerational authority reinforced silence by imposing family "face", prioritizing reputation over truth. The patient's wife was compelled to comply with the patient's father's claim that "he is not sick." This dual power suppression trapped informal caregivers in a paradox: "telling the truth leads to exclusion, telling lies leads to loss of self." Ultimately, this resulted in "narrative fatigue," manifested in extremely low willingness to narrate. In fact, what the informal caregivers recounted was no longer their personal experience, but rather a standardized case history detached from their family identity [11].

2.2.2. Negative impacts

The narrative of silence triggers problems in three areas: individual psychology, medical coordination and family relationship, and it ultimately exacerbates a comprehensive crisis within the informal caregivers' family ecosystem.

First, informal caregivers of patients with rare diseases face significant mental health threats; 86% of caregivers become socially disconnected due to a lack of respite services, and 37% develop depression due to long-term suppression, resulting in physical and mental exhaustion [12,13]. Furthermore, most informal caregivers of patients with rare diseases have reported that they lose personal time due to caregiving, yet fail to gain understanding from various social relationships, thereby endangering their mental health [14]. In this case study, the patient's wife and adult son, who served as informal caregivers, reported that internal family care work became the primary burden outside of daily work and school life. The patient's wife described her time working outside the home as a form of personal relaxation, which briefly freed her from the caregiving burden. Moreover, providing daily care and emotional support for the patient after work became the most exhausting part of the day. The boundary between work and rest blurred due to uninterrupted caregiving, leaving informal caregivers without personal or controllable respite. The physically and mentally exhausting state of caregiving forced "silence" to become the optimal choice for conserving energy.

Second, the narrative of silence exacerbates communication difficulties for the patient's family in medical coordination. To prevent the patient's subjective language from obscuring the description of their condition, informal caregivers often assist in providing more objective accounts of daily care routines and illness situations. Based on long-term daily nursing experience, informal caregivers acquire a comprehensive understanding of the patient's disease state, becoming "experts" with experiential knowledge. However, their silence obscures subtle changes in the patient's condition, resulting in the loss of accurate descriptions of the rare disease patient's status.

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Finally, silence as a compromise contributes to the deterioration of family relationships. In the Chinese family-centric culture, the family constitutes the core unit of narrative, where individual destiny is deeply intertwined with family reputation. Illness is not only a personal physical crisis but also is seen as a source of stigma [15]. Informal caregivers of patients with rare diseases are thereby caught in a dual structural pressure: as caregivers of the nuclear family, they are morally responsible for caregiving and bear the emotional burden of close-range care; while as extended family members, they are also tasked with safeguarding collective family honor. Silence thus emerges as a passive compromise between caregiving responsibility and fear of stigma. However, the silence caused by structural pressures doesn't easily disappear. It forces family members to tolerate each other, eroding family relationships. In the case, the patient's wife refused to remain silent, but older family members pressured her by assuming everyone else was tolerating. The silence of informal caregivers gradually exacerbates a toxic family atmosphere, as no one can express their emotions openly. As emotions accumulate, close relationships between family members are eroded.

Overall, the narratives of silence have multiple negative impacts on rare disease caregivers and their families. Informal caregivers are prone to physical and mental challenges and increasingly feel socially isolated. Silence also obscures the patient's true condition and hinders effective medical communication. Furthermore, individual narratives of silence are often neglected by other family members, and they are unable to be honest and help each other, which exacerbates the crisis of the entire care ecosystem.

3. Influencing factors of the narrative dilemma

The narrative of silence faced by China's informal caregivers of patients with rare diseases is a systemic dilemma produced by the interlocking and gradual intensification of factors operating at multiple levels. This dilemma stems not only from the unique medical and social characteristics of rare diseases themselves but also from the exclusion by mainstream narrative templates, the internalized suppression within Chinese family structures, and the implicit control of social proverbs. These four factors work together from the surface to the core, from the outside in, continuously narrowing the caregivers' expression space, causing their narratives to fall into the dilemma of being silenced, simplified, or distorted, thereby exacerbating their emotional isolation and identity crisis.

3.1. Characteristics of rare diseases

The complex characteristics of rare diseases directly contribute to the narratives of silence among informal caregivers. Firstly, the high misdiagnosis rate and lag in medical knowledge of rare diseases make it difficult for caregivers to find a legitimate position within mainstream medical narratives. Due to the lack of authoritative and timely medical explanations, some caregivers are forced to turn to "spiritual narratives" or folk explanation strategies to compensate for the absence of biomedical discourse; some patients and caregivers may exhibit "resistant compliance" towards the medical system, refusing to use medical terminology; while others directly turn to "spiritual narratives" to resist biomedical narratives, yet further lose external recognition due to deviating from medical discourse [16,17]. Furthermore, the dilemma of informal caregivers "not being able to clearly explain what illness he has" not only reflects the medical system's silence in the face of rare diseases but also reflects the caregivers' vulnerable position in the knowledge-power structure. Research on informal caregivers points out that the narrative dilemma of caregivers is essentially a product of knowledge-power inequality. On the one hand, doctors label caregiver experiences as

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"unscientific observations." On the other hand, caregivers and patients are actually more familiar with the rare disease than doctors are but cannot gain recognition due to barriers in professional terminology. The inversion of knowledge authority within the doctor-patient relationship exacerbates the structural imbalance in the doctor-patient relationship during the treatment of rare diseases. Cultural requirements in different regions also influence caregivers' narrative expression. Italian rare disease caregivers can attempt to use religious metaphors to express pain, but under the influence of China's "face culture" and disease-related shame, informal caregivers fall into the narrative dilemma of "whoever cares will loses face," making it difficult for them to construct an appropriate caregiving role.

Secondly, the highly unknown and complex nature of rare diseases themselves often traps caregivers in a dual dilemma of professional medical tasks and cognitive confusion. Since rare diseases are mostly genetic, chronic, and little-known, and are strongly rooted in uncertain knowledge fields, conflicts over the legitimacy of knowledge often occur in the medical field of rare diseases. More seriously, the medical system's cognitive lag and frequent misdiagnosis regarding these diseases often cause secondary harm. Current medicine still struggles to address the prevention, treatment, and nursing processes of rare diseases. Informal caregivers often need to perform medical tasks that should be executed by professional healthcare staff, such as medication management, rehabilitation training, and even the use of medical instruments. However, studies show that less than 20% of caregivers have received relevant training. The identity of being "unlicensed professionals" exacerbates the physical and mental load and self-doubt of informal caregivers.

Finally, rare diseases frequently face social misunderstandings and public scrutiny, which impose greater psychological pressure on informal caregivers and draw them deeper into the narrative of silence. Some rare diseases manifest with visible symptoms, such as the abnormal posture of patients with spasmodic torticollis, or the facial features associated with certain genetic diseases, rendering patients highly vulnerable to public staring, questioning, and stigmatization in public spaces. Widespread ignorance and moralized associations surrounding rare diseases prevail in society.

In the rare disease family of this study, the particular rarity of the disease exacerbated the difficulty of physiological nursing care. For families with rare diseases in this study, the rarity of the disease exacerbates the challenges of physical care. The wife, as an informal caregiver, not only has to navigate the uncertainty surrounding the disease itself but also undertakes operational tasks that should be the province of medical professionals. This overwhelming workload can make informal caregivers more vulnerable to silence under the immense pressure of caregiving. Furthermore, the medical system's lag in understanding this rare disease and frequent misdiagnoses inflict secondary harm on them. Medical "definitional aphasia" and diagnostic violence not only delay treatment and exacerbate symptoms, but also further undermine the credibility of informal caregivers' words. After repeated unsuccessful medical treatment, they fall into silence and self-doubt, even resorting to unscientific folk remedies, resulting in significant financial and psychological burdens.

Furthermore, the distinctive characteristics of spasmodic torticollis, a rare disease, further increase its visibility and stigma. The social gaze of everyday life views the physiological abnormalities of rare diseases as "abnormal" or "ominous," which not only increases patients' sense of shame but also forces caregivers to adopt avoidance strategies and be reluctant to share their concerns. The widespread social ignorance and stigma surrounding rare diseases places informal caregivers under pressure from external misunderstandings and malicious interpretations, further trapping them in a silent predicament.

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Overall, the intense demands of daily care and doctor-patient coordination lead to narrative fatigue. The medical system's misdiagnosis and neglect of rare diseases undermines the legitimacy of these narratives, and the easily visible symptoms of rare diseases easily lead to painful narratives. This triple pressure makes these informal caregivers more vulnerable to silence.

3.2. Mainstream illness narrative preferences

Disease narratives display specific dominant preferences, which often put pressure on informal caregivers at the societal level, reinforcing their narratives of silence.

Globally, particularly in the Western narrative traditions, illness stories typically privilege the "restitution narrative," where the illness is overcome and the patient recovers [18]. In contrast, the "chaos narrative" is often rejected [19]. However, patients with rare diseases are characterized by long-term chronicity and difficulty in being cured. This makes it hard to fit the "restitution narrative" template. While the narratives, such as the "chaos narrative", which can more truthfully reflect the inexplicable pain and heavy burden informal caregivers face, it is undervalued. Consequently, the narratives of informal caregivers of patients with rare diseases often appear invalid or ineffective.

"Restitution narratives" may generate a narrative of silence. Specifically, the "restitution narrative" is a socially preferred mode of illness story. It demands a positive outcome: the patient ultimately recovers. To meet this expectation, informal caregivers are often compelled to narrate their experiences using positive language. They tend to downplay or conceal the harsh realities of the illness, including invisible labor, financial burden, and emotional exhaustion. More importantly, they must present "recovery" as the story's endpoint to prevent causing discomfort to listeners. However, for rare disease families, this narrative is almost impossible to achieve. The disease is difficult to cure, daily care is repetitive and exhausting, and there is no recoverable outcome to fabricate. Therefore, their narratives do not fit into the existing "restitution narrative" framework, and their true narratives are often overlooked.

Meanwhile, mainstream society favors "restitution narrative" that portray doctors as heroes who triumph over illness, or "quest narrative" that portray patients as inspirational role models. These narrative preferences neglect informal caregivers. Their narratives are either unheard or rewritten, further deepening their sense of isolation.

In contrast, the "chaos narrative" describes daily struggles that lack clear cause-and-effect relationships, have no recovery ending, and are difficult to package as inspirational stories. It authentically reflects the ongoing uncertainty, inexpressible pain, and heavy burden of patients with rare diseases and their caregivers. Similar to many chronic or difficult-to-cure diseases, "quest narratives" or even "chaos narratives" can provide patients and caregivers with a stronger and more lasting cognitive framework. They avoid over-reliance on the future expectation of complete recovery inherent in "restitution narratives" [20]. When mainstream culture is obsessed with "restitution," but rare diseases require long-term or even lifelong management, the stories of helpless informal caregivers may lack cultural resonance or societal recognition.

It is important to note that while "restitution narratives" and "chaos narratives" can explain the narrative of silence among informal caregivers of patients with rare diseases, they cannot fully explain why this problem occurs in China. Under the pressure of Confucian "harmony in human relations", informal caregivers in China are more prone to the sense of shame, and also face more specific naming taboos [21]. Such shame and taboos intensify the narrative pressure, making Chinese informal caregivers of patients with rare diseases more likely to remain silent.

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3.3. Characteristics of Chinese families

In the Chinese context, the structure and relationships within families contribute to the internalization of the narrative of silence. Chinese families are highly cohesive social units that value external image. They typically follow the principle that "family private matters should not be made public." When facing a rare disease, which may be perceived as a "burden" or "misfortune," families tend to manage the issue internally. They share the burden collectively and strive to present a "normal" image to outsiders. While this internal coping mechanism provides an emotional buffer during the initial stages, it also systematically limits opportunities for individuals to express negative emotions or seek external support. This phenomenon often occurs among primary caregivers. Emotions such as exhaustion, anxiety, and fear experienced by informal caregivers are often regarded as threats to family harmony and external image. Consequently, these emotions must be suppressed or hidden.

Intergenerational relationships and power characteristics within Chinese families can further contribute to informal caregivers' silence. In the case study, elders exercised control over economic resources, moral obligations, and emotional influence. This lead the patient's wife and son to remain silent about the condition. For example, after diagnosis, the patient's father orchestrated concealment of the illness within the family. He attempted to apply therapies originally intended for cervical spondylosis to the patient's daily treatment. The patient's wife was required to report on therapy's efficacy weekly. As a result, she was compelled to participate in a narrative performance of gradual recovery, thereby being trapped in silence caused by the concealment of the disease name. Simultaneously, the patient's father simplified the rare disease to a mere neck pain, leading the patient's wife into semantic silence. The elders not only negated the wife's complaints but also transformed individual pain into a familial duty that must be endured. This forced the patient's wife into silence. It also exacerbated the deprivation of her discourse power.

3.4. Social proverbs

The more profound suppression comes from the social proverbs. Unlike Western culture, China's distinctive proverbs system underpins and shapes the narrative dilemma. Scholars have already noted the dialectical relationship between agency and fate embodied in proverbs such as "do all that is humanly possible, then leave it to fate" and scholars believe they help to explain how Chinese families process trauma through the proverb system [22]. Building on this, this study selects the commonly used, family-oriented proverb "every family has its own hard scripture" as the analytical focus, which is similar to "every family has a skeleton in the cupboard". It analyzes how it participates in constructing the narrative of silence within the context of rare disease caregiving.

Within the Chinese proverb system, expressions like "every family has its own hard scripture" "suffering a loss is a blessing", and "endure for a while, and the wind and waves will calm" function through three interrelated mechanisms: they universalize, fatalize, and moralize individual suffering, thereby transforming particular pain into a "common destiny" that every family must silently endure. Superficially, such discourses provide informal caregivers with a cognitive buffer, endowing their endurance with positive cultural meaning and enhancing the family's resilience in adversity. However, they also subtly suppress the legitimacy of articulation. When informal caregivers attempt to express dissatisfaction and exhaustion, the above proverbs often frame their pain as a common experience, denying the particularity of their suffering and its structural causes.

Firstly, in the case study of this research, the patient's wife was often comforted by relatives and friends with phrases like: "In life, which family doesn't have its bumps? "Every family has its own

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hard scripture". Such responses, through words like "every family" and "all," blur the extreme difficulties brought by the rare disease into a kind of universal norm, thereby dissolving its uniqueness and severity at the discursive level. The narrative strategy implied by the proverb indirectly acknowledges the existence of suffering while incorporating it into an acceptable, universal script of family life, forcing the caregivers to accept reality while simultaneously losing the linguistic space to speak openly about their pain.

Furthermore, the metaphor of "chanting scriptures" implies a strong imperative to internalise moral responsibility. Using "chanting scriptures" as a metaphor, it suggests that the caregivers must silently undertake and "cultivate well" this piece of family homework. Their sense of responsibility is even morally judged by whether they can fulfil the family duty through "proper scripture chanting." The cultural pressure within the narrative prompts informal caregivers to transform external structural problems, such as lagging in medical resources and deficiency of external support, into internal moral responsibilities, and to attribute them to insufficient personal effort or cultivation, thus overlooking the institutional and social causes behind them.

Ultimately, this type of proverb is reinforced within the cultural background of Confucian emphasis on "harmony in human relations", further inhibiting the possibility of making the "difficult scripture" public within the family, often making discussions about illness taboo. The system weakens the caregiver's willingness to express pain. When caregivers attempt to talk about their exhaustion or unfair treatment, the proverb "every family has its own hard scripture" usually treats their pain as an immature acceptance of the "universal law," or blames the problem on the caregivers' failure to "cultivate oneself" better. Therefore, these proverbs are not merely comforting; they actually constitute a kind of narrative guiding tool that provides limited psychological resilience while systematically denying the particularity of individual pain and its structural roots. Social proverbs, combined with family pressure, medical neglect, and social discrimination, ultimately trap caregivers in a closed loop of silence.

Overall, the inherent characteristics of rare diseases directly limit the narrative capacity of informal caregivers of patients with rare diseases. The mainstream cultural narrative and the characteristics of Chinese families intertwine, restricting the narrative willingness of informal caregivers of patients with rare diseases within the family. At the same time, the everyday performance of social proverbs suppresses the narrative opportunities of informal caregivers outside the family. Finally, they are completely trapped in the narrative of silence.

4. Recommendations

The narrative of silence faced by informal caregivers of patients with rare diseases in China requires systematic resolution. To achieve this, it is necessary to construct a supportive narrative ecology at multiple levels. This dilemma stems from four main factors: the characteristics of rare diseases themselves, the exclusion of mainstream narrative preferences, the suppression within families, and the deep-seated regulative power of social proverbs. Accordingly, this paper proposes the following four targeted recommendations aimed at addressing the aforementioned influencing factors at their root and helping informal caregivers break the silence.

4.1. Enhance the responsiveness of the healthcare system

The high misdiagnosis rate, cognitive lag, and social stigma associated with rare diseases are the primary reasons that caregivers fall into silence. The healthcare system should take the lead in making changes. First, it should promote the establishment of national-level definitions, diagnosis

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and treatment standards, and referral guidelines for rare diseases [23]. These measures can shorten diagnostic delays and reduce the rate of misdiagnosis. Second, it needs to conduct specialized training on rare diseases within medical institutions to enhance healthcare professionals' ability to recognize diseases, empathetic communication skills, and narrative competence. Particularly in the process of delivering diagnoses, healthcare professionals should provide clear, transparent information and psychological support. Such practices help prevent caregivers' feelings of helplessness from being exacerbated by poor communication. Finally, public-health campaigns should popularise knowledge about rare diseases through public health education, advocate for destigmatizing social concepts, reduce public stares and moral judgment triggered by visible symptoms, and create a more friendly public expression space for caregivers.

4.2. Break the suppression of mainstream narrative templates

To combat the marginalization of real illness experiences under the dominance of the "restitution narrative," diverse narrative forms should be actively recognized and accepted. It is recommended to develop and promote narrative support tools such as online narrative workshops and emotional expression platforms, providing informal caregivers with low-cost, low-burden expressive channels. More importantly, the healthcare sector, the media sector, and the public should accept the circulation of "chaos narratives." Narratives filled with uncertainty, exhaustion, and hopelessness also have value, and should not be displaced by an insistence on "positive energy" or "inspirational stories."

4.3. Build internal and external family support networks

Addressing the depression caused by intergenerational power within Chinese families, it is necessary to jointly build a support system both internally and externally. Within the family, methods such as family therapy and psychological education can be used to encourage open communication about emotions and stress. At the same time, families should abandon some erroneous concepts in traditional concepts and redistribute care responsibilities and voice. Outside the family, a multi-level care resource network should be constructed, breaking through the traditional model of a single primary caregiver. Specifically, through policy guidance, a support system composed of secondary caregivers, community volunteers, and professional health consultants should be developed to provide psychological counseling, economic compensation, and flexible leave policies, to practically reduce the physical and mental burden of primary caregivers.

4.4. Weaken the implicit discipline of the social proverbs

Proverbs such as "every family has its own hard scripture" often obscure the unique and structural roots of individual suffering. Addressing this issue requires cultural change. First, everyone must not ignore the underlying issues and gradually reduce stigma. Second, public education should be used to improve public awareness and encourage compassionate treatment of families with rare diseases. Finally, within rare disease families, family leaders should foster a culture of friendly communication, which can help families overcome the challenges. Through various strategies, society can gradually weaken the oppressive narratives of deeply ingrained cultural jargon that define specific groups.

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5. Conclusion

This study, drawing on existing literature and in-depth case interviews, reveals the dilemma of the narrative of silence and analyzes the factors. This research argues that the narrative of silence can be divided into three specific dilemmas: performative silence, which involves disguising the illness for external social relationships; semantic silence, which involves discourse isolation within the family; and power silence, resulting from doctor-patient relationships and intergenerational dynamics. All three types exert negative impacts on informal caregivers' mental health, patients' healthcare treatment, and family relationships. However, the narrative of silence is not simply the result of insufficient individual willingness or ability; it is a systemic dilemma shaped by the interplay of rare-disease characteristics, dominant illness narrative styles, Chinese family features, and social proverbs. The medical and social characteristics of rare diseases themselves constitute the objective basis for the narrative of silence. The preference of mainstream illness narratives, particularly the promotion of the "restitution narrative" and the rejection of the "chaos narrative," filters out illness experiences that do not align with societal expectations. Meanwhile, the Chinese family structures and social proverbs further suppress the willingness and possibility of authentic expression. These four factors are interconnected and reinforce each other, ultimately trapping informal caregivers in a closed loop of muteness where they struggle to speak and remain unheard.

At the theoretical level, by introducing local socio-cultural characteristics such as family ethics, proverb metaphors, and cultural shame, this study expands the explanatory power of existing narrative medicine theories in cross-cultural contexts, particularly highlighting the shaping role of the "family" concept on caregiver narrative behavior in the Chinese context, offering a new perspective for cross-cultural studies of illness narratives. At the practical level, this study reveals the multiple negative impacts of the narrative of silence on caregiver mental health, patient medical coordination, and family relationship quality, highlighting that breaking the silence promotes not only individual emotional catharsis but also the sustainable functioning of the entire care ecosystem. Accordingly, four recommendations are proposed: a more responsive healthcare system, diversified illness narratives, stronger family-community support networks, and the critical deconstruction of silencing social proverbs. Together, these aim to create a more supportive and expressive environment for informal caregivers across institutional, cultural and social levels.

However, this study has limitations. Firstly, although the in-depth interview based on a single family can provide rich qualitative materials, the generalizability of the conclusions still awaits verification through larger-scale and more diverse samples. Secondly, the discussion on family interaction mechanisms and social proverbs remains preliminary. Future research could combine methods like discourse analysis to further reveal the micro-level mechanisms of the narrative of silence. Furthermore, although this study emphasizes the influence of local cultural factors, it does not extend to in-depth cross-national comparative research. Future work could further explore the similarities and differences in caregiver narrative patterns across different cultural systems.

Looking ahead, research can deepen in three aspects. Theoretically, further dialogue between illness narrative theory and local sociological concepts is needed. Methodologically, more in-depth qualitative research using ethnographic and discourse analysis methods can clarify the pathways shaping informal caregivers' narrative willingness and ability. Practically, new perspectives should track the dynamics of caregiving practices and continuously assess the effectiveness of interventions and policy translation. Only in this way can informal caregivers of patients with rare diseases truly find their voice and break free from the silence.

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