

Study on Social Awareness and Patient Needs of the Rare Disease Epidermolysis Bullosa

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Abstract

This article primarily explores the current situation of the rare disease known as "Epidermolysis Bullosa," the challenges faced by patients, societal awareness, medical insurance conditions, and the status of public welfare activities. Patients face issues such as a shortage of specialized medical personnel, unclear diagnoses, susceptibility to discrimination, poor accessibility to medications, and lack of adequate support from the health care system. Through methods such as text analysis, interviews, and questionnaires, the study provides an in-depth analysis and proposes suggestions like enhancing societal awareness, improving medical insurance policies, and accelerating relevant scientific research to promote social attention and support for rare diseases treatment and improve the quality of life for patients with this disease, who are called "Butterfly Children" because their skin is as fragile as a butterfly's wings.

Keywords

Butterfly Children, Epidermolysis Bullosa, Public welfare, Rare disease.

1. Introduction

Among the vast 20 million rare disease patients in China, there are approximately 1,200 EB patients. Due to their inability to synthesize the structural proteins needed for their skin, their skin can easily blister with the slightest external force, making it as fragile as "butterfly wings," hence they are also known as Butterfly Children.

As rare genetic skin disease patients, Butterfly Children are facing issues such as a shortage of specialized medical personnel, unclear diagnoses, susceptibility to discrimination, poor accessibility to medications and lack of adequate support from the health care system due to the significant urban-rural disparity in the current health care system.

Due to the low prevalence rate and small patient population of the disease, EB treatment projects are not covered by the health care system, and there is limited propagation and policy support on related topics, resulting in minimal impact. On July 2, 2019, the China Red Cross Foundation launched the "Rare Disease Care Action" in Yinchuan, Ningxia. This plan aims to provide public welfare assistance to EB patients through organizing nursing knowledge training, providing dressing change guidance and financial aid for patients in need. In November 2022, the National Medical Insurance Drug List was adjusted to include 45 drugs for 26 rare diseases, including Epidermolysis Bullosa (EB). Regarding the propagation of information about Butterfly Children, there are currently only three WeChat public accounts dedicated to this topic: two are social organization accounts ("Shanghai Debo Butterfly Children Care Center" and "Qiu Ai Tong Xing Charity Foundation"), and the third is a personal account of a patient ("Prince Superman"). The content of the "Prince Superman" account mainly includes interview videos with other EB patients, sharing their experiences with the disease. The audience consists of socially concerned individuals and other rare disease patients. The videos can typically gain tens of thousands of views on the video platform Bilibili, helping more people understand the situation of Butterfly Children and encouraging more Butterfly Children

patients to integrate into public life. However, compared to other general entertainment videos on the internet, their traffic is relatively low, and their impact remains limited.

Butterfly Children often endure unimaginable pain and social discrimination due to their limited mobility or unique symptoms. For example, 44-year-old Gao Chongyi is one of the 112 registered EB patients in Shandong. He was once rejected by schools due to discrimination. "Some schools said I had an infectious disease, and others were afraid of taking responsibility, so they just didn't accept me." It was only through his parents' relentless efforts that he was able to attend school. Difficulty in attending school is a common issue faced by all EB patients. Besides the discrimination caused by misunderstandings, EB patients also suffer from a low quality of life. Currently, there is no effective treatment for this disease. Depending on the severity, the complete process of puncturing blisters, debridement, changing dressings, and bandaging can take 2 to 4 hours. In most families, at least one parent has to resign from their job to take care of the child.

Most Butterfly Children patients' families feel helpless and guilty, while the patients themselves experience anxiety and depression, leading to isolation from society. However, there are some patients who face their pain positively like Wang Zichao, the owner of the "Prince Superman" account. His audience, after watching his videos and reading his articles, see a kind of spiritual strength in him. In 2020, a netizen suffering from a psychological disorder once contemplated ending his life one night, but was inspired and moved by Wang Zichao's story and abandoned the idea of suicide.

2. Literature References

Currently, research and treatment of rare diseases in China are still in the initial stages. There are few institutions and experts studying rare diseases, and most rare disease patients are misdiagnosed or undiagnosed for a long time, unable to receive timely and effective treatment. Although many tertiary-level hospitals have focused on clinical research and treatment of rare diseases, and have preliminarily met the basic conditions for diagnosing and treating rare diseases and conducting scientific research activities, they still find it difficult to meet the needs of these patients in terms of treatment. The small number of patients and their scattered distribution, leading to the low commercial value and the difficulty of clinical trials stem, are the biggest fundamental obstacles to the research of rare diseases and orphan drugs. Given the limited health resources in China, to promote the healthy development of the fund, financial support can be provided for orphan drugs that enter the clinical trial stage, and after their market launch, a batch recovery system can be implemented. Due to the small number of users of orphan drugs, tax reductions and exemptions can be considered for several years after the drug is marketed. Based on the general income and expenditure situation of new drugs, an initial period of 10 years or more can be set, with appropriate adjustments made as needed to ensure that orphan drugs are profitable after being marketed, forming a virtuous cycle.

Most patients have recognized the rarity of their diseases, while a small number of patients have not correctly recognized their diseases as rare diseases. The main reason is that there is currently little publicity about rare diseases in China, and social awareness is low. The general public's awareness of rare diseases is at an even lower level. It is basically very difficult for rare diseases patients to apply for commercial medical insurance. Suffering from rare diseases imposes a great economic burden on individuals and families, easily leading to poverty due to illness. The vast majority of patients believe that the existing health care system still has room for improvement. Most rare disease patients believe that China needs and is fully capable and necessary to establish a special rare disease health care system under the existing system.

Song Hai et al. (2020) believe that clinical physicians have a average understanding of the pathogenesis, incidence, and heredity of rare diseases, and a low understanding of the

transmitting Population. The higher the education level of the physician, the higher their awareness of rare diseases; the shorter the years of practice, the higher the awareness of rare diseases; tertiary hospital physicians have a higher awareness of rare diseases than secondary hospital physicians. Physicians with longer years of practice have lower enthusiasm for learning about rare diseases, a new medical field, due to a lack of competitive pressure and being distracted by various family and social affairs, resulting in a lower level of understanding. Clinical physicians in medium-sized cities have less contact with rare diseases in their daily work, fewer opportunities for academic exchanges, thus very low awareness of rare diseases. Zhang Shu and Wei Ning (2024) proposed that rare disease patients, due to long-term physical pain, have unstable emotional status and exhibit aggressive behaviors; due to their illness, they cannot participate in daily group activities and lack opportunities to interact with peers; the strange looks from others lead to a lower willingness to communicate with people. Patients lack participation in daily group activities, and although they can attend school, they miss important socialization processes and cannot learn social interactions through contact with other peers, making it difficult to integrate into groups. Social workers mainly help patients by alleviating family tensions, reducing caregiving pressure, facilitating school enrollment, establishing peer relationships, and providing medical resources.

3. Social Awareness and Needs Analysis of Butterfly Children

3.1. Findings Based on Text Analysis

This study conducted a search on the WeChat public platform using the keyword "Butterfly Children," collecting ten in-depth reports with a total of 47,000 words, sorted by popularity, for text analysis. Through high-frequency word analysis, it was found that "patient" appeared 95 times, the highest frequency, as Butterfly Children suffer from genetically related conditions. "Mother" appeared 84 times, and "father" appeared 36 times, reflecting that from the time Butterfly Children are diagnosed at birth, they are more often cared for by their mothers, while fathers take on the responsibility of earning money. Due to the high cost of daily medications and the need for dedicated care for Butterfly Children, there is a division of labor in families where fathers work for money and mothers provide care. The term "keep a clear and calm mind" appeared 42 times, indicating that the families of patients actively adjust their mindset, evolving from initial panic or even despair to a relatively peaceful state, hoping to provide a worry-free life for their children. "Pray" appeared 27 times, showing that when families first learn that their child may have a rare disease, their psychological state is one of confusion. Lacking medical information and resources, they can only alleviate their anxiety through prayer. "Fellow patients" appeared 27 times, reflecting that Butterfly Children and their families have a socially connected network through which they exchange information about the condition, caregiving knowledge, and provide mutual support. "School" appeared 20 times, and "study" appeared 19 times, indicating that due to their illness, Butterfly Children may not be able to continue attending school after a period of school life, spending more time at home.

3.2. Findings Based on Interviews

The interviewee, Haohao's mother, stated that her child has had EB since birth in 2011, with no skin on his left leg, and was immediately sent to Beijing Children's Hospital for treatment. A month later, the doctor recommended nursing the patient at home, and currently, the child's health can only be maintained through parental care. The child's condition is very severe, with the skin on his hands and feet frequently ulcerating and sticking together, and no other cases have been reported in Fujian besides Haohao.

Economic difficulties: Haohao's mother works in a factory, and her husband is a Didi driver. Their income is meager, making it difficult to afford the child's medical expenses. They have

tried to raise funds for the child through various channels, but with limited success. To treat the child, they have spent all their savings and borrowed money from relatives. When the child was younger, they received financial support from the public through crowdfunding platforms like Qingsongchou, but they no longer seek help now. Since they live in a rural area, their relatives live nearby and occasionally help take care of the child, such as boiling water and bringing bread, but the primary care still falls on Haohao's mother and father.

Child's psychological issues: Due to the condition, the child is irritable, depressed, and unwilling to communicate with others. Haohao's mother stated that during this period, the child faced discrimination at school. She asked the teachers and principal to take care of him, but the results were not satisfactory, making him more withdrawn. Haohao is a very determined child who loves reading, spending three to four hours a day on it.

Social security situation: Haohao's mother stated that due to the disease, they cannot purchase insurance for the child, and medical security system can only cover part of the medication costs, which undoubtedly adds to the difficulty of the child's treatment.

The interviewee, Sirui's mother, introduced the child's birth date, symptoms at birth in 2016, and diagnosis. The child currently suffers from multiple complications, such as skin damage, vision problems (one eye is blind, and the other has blurred vision), bent hands and feet, esophageal atrophy, and inability to eat even rice. They first went to the county hospital, then the city children's hospital, and finally Fudan Hospital in Shanghai.

Sirui's mother stated that they live in a township in Jiangyin City, with poor family economic conditions. She has resigned to take care of the child at home, and her husband is unemployed. The child has one or two friends but rarely has the opportunity to play with others because the child takes risks of injury and the activity she can participate is limited. The car and house of the family are provided by the grandparents. Other relatives have their own children to take care of, have no time to help, and lack experience in caring for children with this condition.

Child's psychological condition: Sirui's mother mentioned that the child has a bad temper, possibly related to the disease and being spoiled by parents from a young age. The child has the best relationship with the mother in the family and communicates everything with her. The child faces certain restrictions at school, such as not being able to participate in physical education, flag-raising ceremony, and sports events. The child also needs the mother's accompaniment to the bathroom. The mother needs to be on standby at the gate to prevent the child from getting injured.

Social assistance: Sirui's mother mentioned two charity organizations, the Xu Rongxiang Foundation and the Ice Bucket Challenge Foundation, which can reimburse some medical expenses for the child. Sirui's mother mentioned that if she spends 30,000 yuan a year on dressings out of pocket, with a salary of 60,000 yuan, she can get about 2,000 yuan reimbursed. There are Butterfly Children gathering events held in Shanghai, but she has not participated because she takes care of the child alone and cannot stay overnight outside.

The interviewee, Roy, stated that he engaged in some overseas public welfare activities and projects before the pandemic and later focused on some public welfare activities in rural China, encountering some rare disease cases mainly through personal connections.

Propagation of awareness of rare diseases: He focuses on the propagation of awareness about rare diseases in society, hoping to let people know that some diseases are not contagious and reduce discrimination against patients.

Fundraising methods and trust issues: Roy stated that current fundraising methods are based on trust, requiring the public to believe in the use of the funds. He mentioned two methods: one is fundraising within familiar social circles, and the other is fundraising through foundations and the general public.

Medical difficulties for rare disease patients' families: Roy focuses on the medical difficulties faced by rare disease patients' families in rural areas, mentioning that insufficient medical awareness and transportation issues prevent patients from receiving timely treatment.

Publicity Activities and Strategies: Roy stated that they hope to raise awareness about rare diseases among more people. They plan to organize activities that allow the public to experience the lives of patients and distribute promotional materials. He suggested focusing on the details of the activities to enhance participation and propagation effectiveness.

4. Questionnaire Survey

This survey aims to understand respondents' awareness, attitudes, and behavioral tendencies regarding rare diseases and Butterfly Children. Through the questionnaire survey, information was collected on respondents' understanding of rare diseases, their level of concern for Butterfly Children patients, their willingness to participate in related activities, and their emotional reactions to the related content. The survey results will help assess the level of social concern for rare disease patients and provide reference for related charity work.

Firstly, the level of understanding of rare diseases is generally average, with 30.84% of respondents indicating they know a little, and only 3.74% indicating they are fully aware of rare diseases. Secondly, most respondents have a low level of understanding of rare diseases and Butterfly Children, and their evaluation of society's attention and support for rare disease patients is generally low. Additionally, most respondents are unsure whether it is necessary to promote public awareness of Butterfly Children and have not shown enthusiasm for participating in related charity activities. Finally, in terms of emotions, the vast majority of respondents felt sympathy and will to encourage the patients, while a small number felt pity.

5. Literature References

Firstly, Butterfly Children patients are emotionally unstable due to the impact of the disease. The stress caused by the disease makes them more irritable compared to those who are not affected.

Secondly, Butterfly Children patients face discrimination. Most people are not familiar with this disease and may worry about the risk of contagion, leading to discrimination. Additionally, due to the skin ulcerations of Butterfly Children, they may also face discrimination based on their appearance.

Thirdly, activities like the Ice Bucket Challenge gained widespread attention mainly for two reasons: firstly, the challengingness of the activity attracted many people's attention and curiosity; secondly, it was initiated by celebrities, which added to its popularity. Therefore, activities for Butterfly Children can focus primarily on the first point—the challenging of the activity.

6. Conclusion

Nowadays, with the acceleration and diversification of information dissemination, along with various public welfare campaigns and social activities, more people can get a glimpse of the real lives and inner worlds of these minority groups. Take the "Butterfly Children", for example. In the past, their difficult living conditions rarely received public attention. Now, more and more compassionate individuals are advocating for them, sharing their stories of battling illness with resilience. This has led to a growing understanding of their struggles and a willingness to offer support and help. Instead of strange looks and cold indifference, people are approaching them with respect and care. This shift undoubtedly lays a solid foundation for building a more

inclusive and friendly social environment, allowing every minority group to feel the warmth of humanity and the dawn of hope.

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