Rare but Not Alone: The Challenge of Rare Disease Patients and Families in Taiwan

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Rare diseases affect only a small number of people, making drug research and development costly and challenging. Diagnosis is often difficult, clinical trials are hard to conduct, data collection is scarce, and payment hurdles are high. Consequently, few pharmaceutical companies are willing to invest in this area. However, Taiwan has emerged as a global leader in rare disease treatment.

Taiwan is the fifth country in the world to enact legislation protecting patients with rare diseases. The country has introduced several laws, including the "Enforcement Rules of the Rare Disease Prevention and Drug Law", "Rare Disease Medical Subsidy Measures", "Orphan Disease Drug Project Application Measures", and "Incentive Measures for the Supply, Manufacturing, Research and Development of Rare Disease Drugs". These regulations integrate the prevention and treatment of rare diseases with the Orphan Drug Act, making Taiwan a pioneer in this field.

To ensure that rare disease budgets are not compromised within the national health insurance system, Taiwan established a "rare disease special fund" in 2005. This fund allocates a fixed annual amount for the treatment of rare diseases, which was approximately NTD 8.7 billion in 2022[1]. Taiwan's comprehensive approach to rare disease care and treatment has become a model that other countries aspire to follow.

In recent years, major pharmaceutical companies have increasingly invested in the research and development of drugs for rare diseases, earning foreign certifications from authorities such as the U.S. Food and Drug Administration (FDA) and the European Commission. This progress is a significant boon for patients. As of October 2023, the government has recognized 245 types of rare diseases and announced 140 types of orphan drugs. Health insurance covers 76 of these drugs, capable of treating 40 types of rare diseases. Each year, thousands of rare disease patients benefit from these treatments, significantly improving their health, quality of life, and even saving lives[2].

Although rare diseases affect a small number of people, understanding the patient journey is crucial. Through in-depth interviews, we analyze this journey from the patient's perspective to help pharmaceutical companies identify effective treatment directions

In recent years, the concept of "patient centricity" has become increasingly important in understanding patients' needs. Ipsos conducted one-on-one in-depth interviews (IDIs) to understand how doctors and patients in Taiwan view the unmet needs and various burdens faced by rare disease patients. By adopting the "patient journey" perspective, we comprehensively analyzed every stage—from symptom onset and seeking medical assistance to testing, disease diagnosis, treatment goal setting, treatment selection, treatment expectations, treatment effects, living burdens, unmet needs, and future treatment expectations. This thorough exploration highlighted the differences between doctors' and patients' treatment goals and expectations. These analyses provide valuable insights for future medical market research in Taiwan, offering pharmaceutical companies strategic directions to better support the medical needs of rare disease patients.

- **The journey to a diagnosis for rare disease patients is often lengthy, as low awareness causes delays in seeking medical treatment, leading to worsening symptoms**

  Though in-depth interviews with patients, we uncovered the pre-diagnosis journey of rare disease patients. Many initially seek medical help at the onset of symptoms but often receive only symptomatic treatment. The underlying cause and name of the disease remain unknown, leading to diagnostic delays that can span several months or even years.

  For example, patients with generalized Myasthenia Gravis (gMG) and Paroxysmal Nocturnal Hemoglobinuria (PNH) revealed that, based on doctors' past experiences, it typically takes 3-4 months for patients to receive a diagnosis. Some patients, however, endure more than two years of repeated transfers and examinations before receiving a diagnosis.

- **Some paramedics have insufficient awareness of rare diseases, making diagnosis particularly challenging. As a result, some patients may never receive an accurate diagnosis**

  Low awareness of rare diseases is not limited to the general public; even doctors may struggle to effectively identify them. For example, a patient with generalized Myasthenia Gravis (gMG) visited two or three hospitals, consulting ophthalmologists and traditional Chinese medicine practitioners, but it was not until the condition worsened six months later that he was admitted to the emergency department and subsequently referred to neurology for further diagnosis. Similarly, a survey of physicians treating Hypophosphatasia (HPP) revealed that only 70% of specialists who do not primarily treat HPP have even heard of the disease, indicating a significant lack of familiarity. Limited diagnostic methods further complicate the situation, restricting the ability to accurately diagnose these conditions. This not only delays patient diagnosis and treatment, exacerbating symptoms and missing the optimal treatment window, but also leads to an underestimation of the actual number of cases, preventing patients from receiving appropriate drug treatments.

- **Public awareness of rare diseases is low, leading to misunderstandings about patients**

  Patients have pointed out that, beyond limited drug choices and treatment effects, the most critical issue is the discrimination and lack of empathy stemming from the general public’s ignorance about rare diseases. This is an area that urgently needs improvement.

- **Existing drugs offer limited choices and effectiveness, prompting a need for the swift introduction of foreign-approved medications with NHI benefits**

  For doctors, current treatments still have room for improvement regarding effectiveness, side effects, and infection risks. They believe there is a need for more effective drugs and hope that foreign-approved medications will be introduced in Taiwan soon. Additionally, they advocate for expanded health insurance coverage to include a broader range of medications, providing patients with more treatment options.

To better care for rare disease patients, Taiwan's pharmaceutical companies should focus on several key areas: accelerating diagnosis to avoid treatment delays, offering innovative treatments and more drug options, improving public awareness to reduce misunderstandings, and providing greater emotional support. Collaboration across various sectors is essential for delivering better care to domestic patients.

Furthermore, during the drug benefit evaluation stage, it is crucial to give more attention to patient opinions. Currently, Taiwan's healthcare technology assessment (HTA) processes primarily involve payers, the medical community, and relevant officials. Placing the patient at the center and incorporating their voice into drug benefit decisions is vital. This ensures that the perspectives of rare disease patients are heard and integrated into decision-making, which is the future direction for Taiwan's healthcare efforts.

Reference

- Taiwan Foundation for Rare Disease
  https://www.tfrd.org.tw/tfrd/rare_a#

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