The Social Integration Challenges for the Population with Rare Diseases

Yueanshu Fu[1,a,*]

[1]*Canterbury School, New Milford, 06776, United States*
a. yueanshufu@gmail.com
**corresponding author*

Abstract: Rare illnesses affect people of all ages across the world. Patients, families, and caregivers have many unmet medical and social requirements, resulting in significant mental and financial hardships. Efforts to enhance diagnostic skills and create medicines for an estimated 7000 uncommon illnesses have been very successful. In the United States, a rare illness or condition affects less than 200,000 persons. In the European Union (EU), a rare illness is defined as one that affects fewer than five persons in every 10,000. However, no effective medicines exist for 90% of uncommon disorders. There is a need to increase awareness, advocacy, and outreach to everyone, especially those with low incomes, low literacy, minority ethnic status, and living in underserved and marginalized communities in urban and rural regions, as well as developing countries throughout the globe. The acceptance of patients as research partners complements greater research focus and large regulatory activities, which contribute to faster review and approval programs for goods for severe or life-threatening disorders. The social challenges and lack of recognition of these illnesses cause the patients and their families facing the server difficulties when they try to get treatments, job opportunities, or engage in society. This paper aims to analyze the problem with various research and resources, which seeks to raise awareness of the rare disease population and bring more benefits and help to them.

Keywords: Social challenges, rare diseases, global recognition, orphan.

1. Introduction

A rare illness is defined by a limited number of individuals. Over 7000 rare illnesses afflict more than 30 million individuals in the United States, with each affecting less than 200,000. Furthermore, for the 10% of persons with uncommon diseases and their families, these circumstances are no longer unusual [1]. Molecular genetics has revealed the etiology of several rare ailments, providing new opportunities for patient identification, phenotyping, and treatment development.

Rare illness research presents investigators with unique obstacles that need specialized methods to: (1) clinical study design; (2) research program financing; (3) the development, testing, and approval of novel medications; and (4) clinical scientist training. Rigorous, statistically valid, natural history-controlled, cross-over and n-of-1 trials may demonstrate effectiveness and support regulatory approval of innovative rare illness medicines.

The United States Food and Drug Administration's Orphan Drug Act has encouraged companies to invest in clinical studies to find treatments for uncommon illnesses. For trainees motivated to find

a cure for a rare ailment, dedication to patient longitudinal care offers a foundation for phenotypic and natural history characterization, a source of inspiration, a target population for research, and financial support [1]. The World Health Organization has recognized more than 5000 disorders that fit this criterion. As a result, the projected number of people affected by a rare disease in Europe is more than 30 million, while in North America it is over 25 million [2-5]. The bulk of these disorders lack epidemiological data, making it difficult to accurately determine their prevalence in Europe and other locations.

2. Definition

The term "rare diseases" refers to an extensive range of diseases that tend to influence all physiological systems. The majority of uncommon diseases are caused by genetic defects that result in substantial disability, shortened life expectancy, and decreased physical and mental skills. These constraints reduce an individual's overall quality of life and affect their capacity to pursue education and earn a livelihood. Inborn metabolic mistakes are one example of this, with the bulk of them having a low recurrence rate [6]. A prospective study done in Italy between 1985 and 1997 focused on patients aged 0 to 17. According to the research, just 11% of the 1935 newborn babies identified with inborn metabolic abnormalities survived to maturity [7].

Uncommon diseases may cause tremendous difficulty for the families affected by them. To assess this burden, a questionnaire was sent to 2500 patients suffering from chronic diseases, with 8.2% classed as rare ailments [7]. Individuals suffering from rare diseases faced the greatest adversity in terms of social and economic opportunities, as well as access to medical treatment.

As human, people are living together as a group, which forms the structures of society. People with uncommon illnesses may face long diagnostic journeys, limited treatment choices, and little awareness of their problems. Health disparities continue to be a significant barrier to treatment for persons living with rare diseases, and they must be addressed as a primary priority.

There are about 10,000 uncommon illnesses, with more than 90% without an authorized therapy. On average, it takes almost five years—and frequently longer—to acquire a rare illness diagnosis, with several physicians, experts, and misdiagnoses along the way [4]. Many uncommon illnesses are difficult to identify, making misdiagnosis likely, especially neurological disorders or diseases that appear in infancy or adolescence.

3. Challenges Accompanied with Rare Diseases

Living with a rare ailment, both before and after diagnosis, may complicate almost every area of life. People with uncommon illnesses, together with their caregivers, may face economic hardship, increased social isolation, major treatment delays, and inferior health outcomes. People with uncommon illnesses face much greater obstacles in nations with inadequate medical, social, and economic infrastructure.

The physical conditions of people with rare diseases become a barrier for them to involve in society because people prefer to notice the differences between one another before they to take the time to learn what is inside of them. This situation creates numerous challenges for the people with rare diseases to find jobs, make friends, to live like normal people do.

Rare diseases pose a serious public health risk and a challenge to the medical profession. They are referred to as health orphans since uncommon ailments were overlooked for many years. Similarly, before the United States passed the Orphan Drug Act in 1983, the pharmaceutical industry overlooked the development of medicines for rare diseases, giving rise to the term "orphan pharmaceuticals." The 1989 report of the United States Government's National Commission on Orphan Disease raised public awareness of the challenges that persons with rare diseases experience [6]. The Commission's

hearings with hundreds of stakeholders highlighted issues affecting patient care, such as a lack of information on rare diseases, funding challenges, the drawbacks of providing adequate health insurance and medical expense coverage, and a scarcity of effective therapies.

The International Classification of ailments (ICD) used by the majority of countries makes it difficult to diagnose atypical ailments [7]. The absence of a universally accepted coding system impedes proper patient registration in national or international databases, making it hard to evaluate the economic and social effects of rare diseases. Researchers, patient groups, governmental entities, and pharmaceutical businesses have all developed and maintained national or global registries for a variety of illnesses. The European Rare Illness Task Force, part of the European Commission's Health and Consumers Protection Directorate General, has organized a working group to communicate with WHO on ICD-10 and is assessing all other existing categories in order to build a single framework for the rare illness community (Table 1) [8].

Table 1: Rare diseases with the highest estimated prevalence.

	Estimated prevalence (per 100 000)		Estimated prevalence (per 100 000)
Brugada syndrome	50	Non-Hodgkin malignant lymphoma	30
Erythropoietic protoporphyria	50	Retinitis pigmentosa	27·5
Guillain-Barré syndrome	47	Gelineau's disease	26
Familial melanoma	46	Multiple myeloma	26
Autism, genetic types	45	α1 antitrypsin deficiency	25
Tetralogy of Fallot	45	Congenital diaphragmatic hernia	25
Scleroderma	42	Juvenile idiopathic arthritis	25
Great vessels transposition	32·5	Neurofibromatosis type 1	25
Focal dystonia	30	Oesophageal atresia	25
Marfan's syndrome	30	Polycythaemia vera	25

4. Global Measures and Organizations for Rare Diseases

The European Organization for Rare Illnesses (Eurordis) and Orphanet worked with the European Commission to analyze the frequency of rare illnesses [7]. This study not only evaluated the prevalence of various unusual diseases (see table), but it also indicated a lack of precise data, low consistency among sources of information, and poor methodological quality in epidemiological studies. Furthermore, facilities for biochemical or genetic testing are few.

There are several common conditions that people with rare diseases when they trying to be involved in social activities [8]. First, parents and caregivers struggle to locate schools that are willing and able to accommodate children with uncommon diseases. Second, children with unusual or undiagnosed diseases attend community schools around the nation. Every student, even those with unusual diseases, deserves to have a positive school experience. Rare illnesses may have varying effects on children's IQ, behavior, physical conditions, and educational demands. Lastly, the additional expenditures associated with care for uncommon disorders impoverish families, while caregivers and people living with rare diseases struggle to find and keep good jobs.

The nature of uncommon illnesses creates numerous uncertainties, including the accompanying expenses. There is growing evidence that illness expenses stem not just from healthcare usage but also from non-healthcare or indirect sources, resulting in financial hardship. As a result, identifying the locations with the highest socioeconomic burdens is critical for optimal resource planning and allocation. Considering rare illnesses as a whole allows for resource usage comparisons within and across disease groups, as well as the monitoring of total disease burden over time. We examined the PubMed database from its beginning to December 21, 2021, for relevant papers assessing the societal cost of uncommon illnesses. The words "rare diseases" and "financial hardship" or "socio-economic" or "catastrophic health expenditure" or "impoverishing health expenditure" and "cost of illness" were used. The majority of the research only looked at the expenditures of one uncommon illness or the health-care system. Only one set of research in Europe examined the social costs of ten specific uncommon illnesses. A Google search for the keyword "burden of rare diseases" yielded additional research conducted by a private firm in the United States. Only two research tried to assess catastrophic or impoverishing health expenditure (IHE) in the uncommon illness group. No prior research that assessed both cost and financial stress variables in the same cohort [9].

Due to a lack of public knowledge, people living with rare diseases and their struggles are often unseen and unrecognized. Around the globe, around 300 million people live with a rare illness, with 70% of cases beginning in childhood. Most uncommon illnesses are incapacitating, if not fatal. According to research, 57.5% to 65% of uncommon disorders relate to a lower life expectancy. The great majority of uncommon illnesses have a genetic basis, with well-known examples being cystic fibrosis, sickle cell disease, Huntington's, and numerous muscular atrophies and dystrophies. While these instances have been well researched, many more uncommon illness kinds are much less well understood.

With a noticeable dearth of scientific material accessible, the path to acquiring an orphan illness diagnosis may be lengthy and complex. There are 7,000 identified uncommon illnesses, the majority of which non-specialist healthcare workers have never seen before. This often puts patients in awkward circumstances where they are shuffled between physicians until an accurate diagnosis is made. The procedure takes four to nine years on average in high-income nations, which contributes to the sad truth that up to half of Europe's rare illness patients are now unidentified.

The European Platform for Patients' Organizations, Science, and Industry is a cooperation of patients, industry, and scientists founded in 1994 to share information and debate health-care promotion strategies. Its major purpose is to build a powerful European coalition of patient organizations, academics and physicians, and industry, with the discovery of novel medications for rare illnesses being among its top priorities [10,11].

The NORD logo depicts a person gently rising from darkness into light. This is precisely what has occurred with uncommon illnesses during the last decade. Patients with one of these disorders have raised their concerns, which are now firmly on the agendas of health-care professionals, public-health authorities, and policymakers deciding future medical research spending.

Indeed, during the last two decades, public awareness of rare illnesses has grown steadily as a consequence of the efforts of active advocacy organizations comprised of academics (clinical and

basic researchers) and politicians. Furthermore, the unmet demands of people with rare illnesses offer new investment opportunities, as seen by the pharmaceutical industry's interest. However, almost all uncommon illnesses remain incurable [12]. The breakthroughs in our understanding of disease processes, as well as the explosion of information in genetic medicine, signal that it is time for a leap ahead. This goal requires an increase in governmental research programs for rare illnesses, as well as a surge in medication development to treat many afflicted individuals via public and private efforts.

5. Suggestions

There are several things that people may do to assist increased awareness of rare illnesses. Here are some suggestions. First, use social media to raise awareness about uncommon illnesses: Share information about the sickness you or a loved one are suffering from and how it impacts your life. This will help people appreciate the importance of rare illnesses and the need for further research and assistance. Second, attend Rising-Awareness Activities: Various activities throughout the year aim to raise awareness about rare illnesses. One of the most prominent is Rare Disease Day, which is held on the final day of February each year. Participating in events like this may help increase awareness about rare illnesses while also showing support for individuals afflicted.

Moreover, it is crucial to join a patient organization. The National Organization for Rare Disorders (NORD) provides valuable resources for people and families impacted by rare illnesses. Joining a patient organization may help you connect with people who share your experiences and give opportunity to participate in advocacy and awareness-raising initiatives. The government should advocate for more research and financing for rare illnesses: since they generally get less funding than more prevalent diseases, resulting in delayed diagnosis and treatment. Therefore, lobbying for greater research and funding for uncommon illnesses is essential. Lastly, it is crucial to educate the society about rare illnesses. Learning about these disorders can allow you to better appreciate the perspectives of those afflicted, as well as how the individual can help raise awareness and fund research efforts. Every individual may also share the expertise with others to assist them raise awareness and understanding of rare illnesses.

6. Conclusion

Rare diseases impact a large number of people globally, creating physical, emotional, and financial difficulties for sufferers, their families, and caretakers. While attempts to enhance diagnostic skills and create treatments for uncommon illnesses have been fruitful, the bulk of these ailments continue to lack appropriate treatments. There is a need for increased awareness, advocacy, and outreach, especially among low-income, low-literacy, minority ethnic, and marginalized populations. Accepting patients as research partners, as well as implementing regulatory efforts, may help to speed up therapy evaluation and approval procedures. Individuals with rare illnesses experience social obstacles and a lack of recognition, making it difficult for them to receive medicines, career opportunities, and fully engage in society.

Living with a rare illness may affect many parts of your life, both before and after diagnosis. Individuals with rare illnesses and their caretakers may experience financial hardship, social isolation, treatment delays, and poor health outcomes. The physical issues connected with uncommon illnesses may also limit people's capacity to engage in society and obtain work. Rare illnesses have been disregarded for many years, resulting in a lack of knowledge, financial issues, insufficient health insurance coverage, and a paucity of effective medications. The lack of a globally approved categorization system for uncommon illnesses hampers patient registration and assessing the economic and social burden of these disorders. Global initiatives and organizations, such as the European Organization for Rare illnesses and the European Platform for Patients' Organizations,

Science, and Industry, seek to solve these issues and enhance the lives of people living with rare illnesses. To properly assist this group, further research, pharmaceutical development, and budget allocation are still required

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