The Burden of Rarity: Rare Diseases and Future Perspectives

Dear Editor,

A disease is considered rare disease (RD) when it affects less than one in every 2000 people.[1] The World Health Organization defines RD as a debilitating disease or lifelong condition with a prevalence of one or less per 1000 people. However, several developed and developing countries have their own definitions. It is a health condition that affects a smaller size of the individual patient population as compared to other prevalent diseases in the general population and has a startling social cost. The most common RDs include autoimmune diseases and lysosomal storage disorders, such as Pompe disease, Hirschsprung's disease, Gaucher's disease, cystic fibrosis, hemangiomas, and specific types of muscular dystrophies. The most common RDs that affect children are hemophilia, thalassemia, sickle cell anemia, and primary immunodeficiency.

RDs are often chronic, debilitating, and life-threatening in nature and can develop at any stage of life. Few of these diseases are preventable and can lead to unfavorable living conditions.[2,3] Less than five to seven individuals out of 10,000 are affected by RDs, although 6%–8% of the world's population is affected globally.[4] According to the Ministry of Health and Family Welfare, 72–96 million Indians are affected by RDs.[5] According to Orphanet data, among the 6172 unique RDs, 71.9% were genetic and 69.9% were disproportionately impacting children.[6] The scientific knowledge and medical understanding of RDs is inadequate due to their complexity, heterogeneity, and ever-evolving field. RD awareness, as well as a lack of treatment alternatives, may make the entire process from diagnosis to therapy difficult and uncertain. RDs have emerged as a primary public health priority owing to the challenges imposed by their low prevalence, especially their continuous, life-threatening nature and lack of information and expertise.[7]

There is inadequate epidemiological data to quantify the burden, and research and development options are limited. It is difficult to estimate the exact number of people affected by RDs, as the majority of RDs are not documented, except in certain developed countries. Government indecision on the provision of health-care access is caused by gaps in the maintenance of electronic health records or registers of cases noticed in the underprivileged and vulnerable sectors of society. Clinical trials are a crucial component of the drug discovery process. It is completed in stages and involves identifying numerous individuals suffering from particular medical conditions and developing a robust trial design to demonstrate the efficacy of the medicine. This issue significantly worsened in the case of RDs. Finding patients qualified for clinical trials typically involves significant search expenses. Given the rarity of patients and in their entire professional careers, a significant percentage of doctors in practice reported never having seen a patient with a RD,[8] finding the correct diagnosis takes a substantial amount of time. Clinical trials are expected to be expensive because pharmaceutical companies will most likely have to spend a substantial amount of money.

RDs affect not only the individuals who suffer from them but also their families and communities. Due to the prohibitively high cost of treatment, the impact on families is frequently disastrous in terms of both the emotional and financial tolls. Often, the patients' and their caregivers' medical and social needs are unmet, which leads to experiencing a severe psychosocial burden.[9] The individuals with RDs and their family caregivers' vocational aspects (education and employment) will be adversely affected due to the difficulty in accessing health-care medical services for the management of RDs, which leads to poor health outcomes.

A qualitative study from the USA reported that individuals with RDs had experienced three types of stigma: Structurally enacted, interpersonally enacted, and felt stigma.[10] There is a need to enhance the awareness, advocacy, and implementation of outreach programs about these RDs in low-income countries, among marginalized, poor literacy populations. However, research on the impact of RDs on psychosocial, economic, and vocational aspects of individuals with RDs and their family caregivers globally is lacking. Individuals with RD's cannot afford health-care services due to the high cost of treatment, diagnostic evaluations, medications, etc., especially in developing countries.

Usually, healthcare end-up spending is spent on people who are diagnosed late. This may often result in a terminal patient who needs to be managed with expensive therapies and rehabilitation. The government can implement the early detection programs, as it can save time and resources and reduce costs. A patient can be managed with less costly interventions while providing better care and quality of life. Early screening and diagnosis of RDs are critical for the prevention of perinatal and neonatal disorders such as Angelman syndrome, Carpenter syndrome, craniofacial disorder, Hutchinson-Gilford progeria syndrome, and Wiedemann-Rautenstrauch syndrome. In addition, drug development can be performed using an artificial intelligence algorithm that can potentially reduce costs and preclinical work compared to a fraction of traditional methods. The electronic health records of patients with RDs should be should be introduced and maintained so as to aid in future researches on RDs. From this data, crucial health information related to RD patterns can be identified regionally.

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One of the best methods for a proactive approach to therapy is networking and the exchange of experiences. Networking is one of the most valuable assets for people with RDs. Having a community will not only be a poignant reminder of the energy that people with RDs put into their lives every day but it will also be a valuable opportunity for them to feel less alienated in their ongoing struggles.

In summary, the fact that effective therapies are frequently unavailable adds to the amount of agony and suffering experienced by patients and their families. The government must establish a plan to raise awareness of the severity and impact of RDs on patients and their families, and simultaneously conduct research and plan for better diagnosis and treatment, develop pharmaceuticals drugs that are accessible, affordable, and provide insurance coverage for treatment. Furthermore, patients with RDs typically face challenges owing to their low prevalence and lack of knowledge among their healthcare providers. As there is an urgent need to bridge the gap in physicians' understanding of RDs, it is prudent to introduce additional courses on these diseases in the medical curriculum and postgraduate training for physicians. The internet is the primary source of information on RDs, and e-learning programs and courses should be implemented for all medical practitioners.

Patient informed consent

Patient informed consent was obtained.

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