Invisible illnesses: Shining a light on the psychosocial impact of rare diseases

Komal Saini
Chandigarh College of Pharmacy, Chandigarh Group of Colleges, Landran, Mohali, Punjab, 140307
Email: komalsainiks@gmail.com

Abstract
Rare diseases (RDs) impact a small proportion of the population yet can be very debilitating and dangerous. People with RDs suffer unique and substantial obstacles as a result of the infrequent nature of their medical diseases, such as a lengthy diagnostic process, insufficient clinical care, and restricted access to appropriate medications. Historically, patient groups have been the primary drivers behind increasing awareness about these diseases and fighting for government supportive legislation. The burden of RD on patients, caregivers and families, healthcare systems, and society as a whole deserves more exposure and appreciation. Taken together, scientific, social, ethical, and political responsibilities call for more RD inclusion and integration in research and policies, thereby advancing the goal of the UN Resolution to "leave no one behind."

1. Introduction
Invisible illnesses, particularly rare diseases (RDs), pose distinct challenges not just because of their clinical symptoms, but also because of the psychological effect that they have. These illnesses are frequently not well-known in popular culture, which makes it challenging for those who are impacted by them to get sympathy and assistance from the community. The sense of loneliness is a major component of the psychosocial effects of rare illnesses. People with these disorders may find it difficult to get empathy and support from friends, family, and even medical experts because these conditions are frequently poorly understood by others. The already difficult experience of managing a rare illness can be made worse by this sense of isolation, which can result in emotions of loneliness, sadness, and anxiety (1), as mentioned figure 1.

Physiological effects	Social implications	Functional impact
• Low mood • Frustration • Anxiety • Fear • Uncertainty for future • Irritability • Difficulty in making decisions • Restlessness • Muscle tension • Changes in eating habits	• Difficulties in explaining the symptoms to close friends and family • Difficulty in justifying to not present at workplace and school • Isolation due to difficulty of finding other people with the same diagnosis • Lack of psychological care • Difficulty in planning everyday life • Difficulty accessing resources or benefits • Financial difficulties due to the disease	• Loss of independence due to the disease • Loss of abilities due to the disease • Loss of work or academic opportunities due to the disease

Figure 1. Psychological impact on people affected by RDs

Children, adolescents, and their families are rarely affected by RDs, but their impact is vast. US National Institutes of Health characterises such illnesses as occurring once in 200,000 or less, criteria differ worldwide (2). The US definition is tied to the 1983 Orphan Drug Act, which was enacted to incentivize the pharmaceutical sector to study and develop novel medicines for rare diseases (3). In the United States, these ailments are known as "orphan diseases" because the law was known as the Orphan Drug Act (4). More common orphan and RDs include cystic fibrosis, Lou Gehrig's disease, and Tourette's syndrome; less common ones include Duncan's syndrome, Madelung's illness, and acromegaly/gigantism. Often, the prevalence of RDs is an estimate that is subject to change over time. Due to the complexity and common misinterpretation of these conditions, many people find it more challenging to be properly diagnosed or receive the right therapies. Effective therapies ought to be put into place as soon as the ailment has been discovered. Effective targeted treatments that can change the course of the disease and greatly enhance patient and family outcomes and quality of life are only available for 5% of RDs (5). Regretfully, orphan drugs are not only rare but also quite expensive, and their accessibility varies greatly between and within nations (6). The availability of medicines for children is quite restricted: even the pharmaceuticals that are currently on the market are often not designed with children in mind, and the necessary clinical trials to assess their efficacy and safety in this demographic have not been carried out (7). For both common and uncommon disorders, the same strategy applies: we need to understand the underlying mechanisms of the illness, pinpoint the biological targets that can be targeted with medication, and develop new therapeutics. However, the very first step towards the development of medicines—elucidation of pathological mechanisms—has been taken in only about 1000 of all (up to 8000) rare diseases (8). Additionally, the necessary research infrastructures—such as biobanks for most illnesses and patient registries for studies on the natural course of diseases or cohort collections for clinical investigations—are not available. As a result, specific treatments for the majority of the other 7000 RDs are not even on the horizon, and certain RDs appear to be "undruggable" (such as chromosomal diseases) (9).

2. Challenges with RDs

In India, not only is the general populace unaware about RDs, but so are healthcare providers and policymakers. Compared to developed countries such as the US, India is about 40 years behind in realising the need of combating RDs. In developing countries, much of the effort to combat such diseases is concentrated on obtaining orphan medications. There has been little focus on building registries, genetic screening, access to therapy, (10) health insurance coverage, and public perception of these disorders.

India must adopt a decentralised approach to RDs, using epidemiological data to inform policy and legislation. Such policies should allow for the capture and sharing of electronic health data while also protecting individuals' privacy. Evidence from advanced economies shows that patients struggling with RDs are nearly 90% more likely to share their data for further research than others. India should also aim to create, protect, and promote such recording practices (11). Policies should accelerate scientific innovation and advanced clinical research while also allowing key stakeholders such as patients, doctors and carers, and the pharmaceutical sector to connect more easily.

The National Digital Health Mission (NDHM) can aid in the diagnosis of RDs. Various cutting-edge diagnostics were created during the COVID-19 pandemic and can be utilised for RDs. However, Indian specialists feel that scaling these capabilities to the local level will be challenging, even though the country's bio-community already uses them extensively. Traditional views must alter for therapies to evolve and embrace modern technological advancements in a variety of ways (12). Most RDs necessitate ongoing therapy and assistance for the patient to maintain a

reasonable quality of life. Caregiving depletes families physically, emotionally, and financially. As a result, palliative care, rehabilitation, and psychotherapy services must be included as part of the RD management process. Significant resources are necessary, as well as strong government support. It can be challenging for countries like India to shift resources from more common general diseases to RDs. However, reduced occurrence should not be seen as less significant. Implementing the NPRD is merely one step towards resolving the issues of RD management (12).

3. Future directions

RD patient organisations can promote policy adoption and help coordinate treatment (13). Governments ought to use in-depth conversations and patient advocate focus groups to increase public awareness of the needs of RD populations. On one side, patient groups for RDs are essential in promoting patient rights and opportunities for research. On the other side, patient organisations provide the basis for patients' and their families' psychological assistance. Leading the effort were RDs International, EURORDIS (rare disease-Europe), and the Committee on NGOs for RDs, which resulted in the recently passed historic UN Resolution on RDs. It advocates for greater integration and gives the RD population priority on the UN agenda, which represents a dramatic change in the global policy landscape (14). This international campaign brings attention to the needs of the RD community, which makes it possible to develop critical plans and strategies for providing affordable and easily accessible care. RD patient associations can empower patients as well as care takers by being the voice of the RD population, raising awareness and educating the public at the same time. By shedding light on the psychosocial burden of rare diseases and collaborating to raise awareness and support, we can meet the needs of individuals and families affected by these often-ignored conditions.

The last ten years have seen tremendous advancements in RD research, which have fundamentally altered the worldwide policy environment. Among these advancements are the three 10-year goals set forth by International Rare Disease Consortium (IRDiRC) and the UN Resolution that was approved in December 2021 highlighting the need of integrating the RD population in the UN 2030 agenda. Prior research has emphasised about the significance of an early diagnosis as well as the substantial effects of RDs on patients' socioeconomic burden and quality of life (15-16). It is critical to recognise that a lack of medical understanding, a lack of social support system, and a lack of public awareness of RDs are the primary causes of the physical, social, and economic effects of RDs. As RD becomes a worldwide public health concern, RD policies and initiatives in the areas of healthcare, social care, insurance, education, and many more are required to provide a more fair and inclusive community for the RD population. Future research and analysis on RD will probably advance our understanding of all diseases. Here, we suggest that RDs adopt patient-centered, multidisciplinary action plans in the future, with an emphasis on the execution of educational and training initiatives, the abolition of stigmatisation and discrimination, and the formation of an international coalition among multidisciplinary stakeholders (14).

4. Conclusion

In conclusion, RDs have profound negative impacts on patients, their families, and societies, and RD patients often face many challenges to have their needs met. RDs are only recently and progressively becoming a policy priority for the UN and WHO, extending an invitation to increase health policies and initiatives at the regional and country levels. The creation of mobile applications (apps), wearable technology, and remote sensors will all help with the ongoing patient monitoring process to ensure the efficacy and safety of both licenced and experimental drugs. Access to knowledgeable physicians who possess a deeper comprehension of specific uncommon diseases could be facilitated by telehealth and telemedicine (17). Taken together, scientific, social, ethical, and political responsibilities call for more RD inclusion and integration in research and policies, thereby advancing the goal of the UN Resolution to "leave no one behind."

References

1. Rezaei F, Sanagoo A, Peyrovi H, Jouybari L. Persistent suffering: Living experiences of patients with rare disease: An interpretative phenomenological study. Journal of Education and Health Promotion. 2023;12(1):224.
2. Richter T, Nestler-Parr S, Babela R, Khan ZM, Tesoro T, Molsen E, Hughes DA. Rare disease terminology and definitions—a systematic global review: report of the ISPOR rare disease special interest group. Value in health. 2015;18(6):906-14.
3. Thomas S, Caplan A. The orphan drug act revisited. Jama. 2019;321(9):833-4.
4. (https://www.eurordis.org, viewed on April 26, 2024).
5. Fermaglich LJ, Miller KL. A comprehensive study of the rare diseases and conditions targeted by orphan drug designations and approvals over the forty years of the Orphan Drug Act. Orphanet Journal of Rare Diseases. 2023;18(1):163.
6. Degtiar I. A review of international coverage and pricing strategies for personalized medicine and orphan drugs. Health Policy. 2017;121(12):1240-8.
7. Joint evaluation of regulation (EC) No 1901/2006 of the European Parliament and of the Council of 12 December 2006 on medicinal products for paediatric use and regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products {SEC(2020) 291 SWD. 2020. p. 164.
8. Tambuyzer E, Vandendriessche B, Austin CP, Brooks PJ, Larsson K, Miller Needleman KI, Valentine J, Davies K, Groft SC, Preti R, Oprea TI. Therapies for rare diseases: therapeutic modalities, progress and challenges ahead. Nature Reviews Drug Discovery. 2020;19(2):93-111.
9. Julkowska D, Austin CP, Cutillo CM, Gancberg D, Hager C, Halftermeyer J, Jonker AH, Lau LP, Norstedt I, Rath A, Schuster R. The importance of international collaboration for rare diseases research: a European perspective. Gene therapy. 2017;24(9):562-71.
10. Angural A, Spolia A, Mahajan A, Verma V, Sharma A, Kumar P, Dhar MK, Pandita KK, Rai E, Sharma S. understanding rare genetic diseases in low resource regions like Jammu and Kashmir–India. Frontiers in genetics. 2020;11:415.
11. Belzer LT, Wright SM, Goodwin EJ, Singh MN, Carter BS. Psychosocial considerations for the child with rare disease: a review with recommendations and calls to action. Children. 2022;9(7):933.
12. Pingali V, Das N. Rare diseases require support too. Vikalpa. 2021;46(2):129-34.
13. Dharssi S, Wong-Rieger D, Harold M, Terry S. Review of 11 national policies for rare diseases in the context of key patient needs. Orphanet journal of rare diseases. 2017;12:1-3.
14. Chung CC, Hong Kong Genome Project, Chu AT, Chung BH. Rare disease emerging as a global public health priority. Frontiers in public health. 2022;10:1028545.
15. Austin CP, Cutillo CM, Lau LP, Jonker AH, Rath A, Julkowska D, Thomson D, Terry SF, de Montleau B, Ardigò D, Hivert V. Future of rare diseases research 2017–2027: an IRDiRC perspective. Clinical and translational science. 2018;11(1):21.
16. Cutillo CM, Austin CP, Groft SC. A global approach to rare diseases research and orphan products development: the international rare diseases research consortium (IRDiRC). Rare Diseases Epidemiology: Update and Overview. 2017:349-69.
17. Adachi T, El-Hattab AW, Jain R, Nogales Crespo KA, Quirland Lazo CI, Scarpa M, Summar M, Wattanasirichaigoon D. Enhancing equitable access to rare disease diagnosis and treatment around the world: a review of evidence, policies, and challenges. International journal of environmental research and public health. 2023;20(6):4732.