

Roll

Call

Patient registries: the key to accelerating rare disease research

■ By Nisha Venugopal, PhD

A *patient registry* has been defined as an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves a predetermined scientific, clinical, or policy purpose(s). Although used interchangeably with the term “patient registries,” *natural history studies* are more specifically designed as tracking and monitoring the progression of a particular disease over time in a comprehensive manner allowing correlations with genetic, demographic, environmental, and other variables.¹

Both patient registries and natural history studies are important repositories of patient health, diagnosis, and disease progression, and serve a variety of purposes, including facilitating research: basic research, to learn about a disease or condition, and translational research,

for the development of therapeutics and to study and improve the quality and best practices in healthcare and treatment. Patient registries are especially valuable for rare diseases, which suffer from a dearth of information due to low prevalence and a lack of robust methods for diagnosis and standard of care.

The World Health Organization (WHO) defines rare diseases as debilitating, lifelong disorders with a prevalence of less than 1 per 1,000 persons.² The definition varies by country. India is home to an estimated 70 million people affected by a rare disease, per the Indian Council of Medical Research, but currently does not have a definition for what constitutes a rare disease.² The reason cited is the lack of reliable epidemiological data. For the purpose of regulatory approvals, the Central Drugs Standard Control Organization (CDSCO) considers any drug intended to treat a condition affecting fewer than 500,000 Indians as an orphan drug.

There are almost 11,000 rare diseases that have been identified as of today, and more are added to the list daily.³ This list includes autoimmune disorders, congenital malformations, inherited cancers, and a few endemic infectious diseases with low prevalence. Most rare diseases are genetic in origin and many are life-threatening or severely debilitating. Of these, only around 450 have been reported from India.⁴

Patients with rare diseases face obstacles in areas such as access to accurate diagnosis, appropriate treatment options, and clinical trials, which are often the only source of hope for patients suffering from about 95% of rare diseases with no approved treatment. Research on rare diseases presents unique challenges because of their low prevalence. The limited patient population makes it difficult to conduct robust clinical trials, gather sufficient data, and develop effective treatments. This challenge



is exacerbated by the diagnostic odyssey—delayed and inaccurate diagnoses due to the lack of awareness and knowledge about rare diseases.

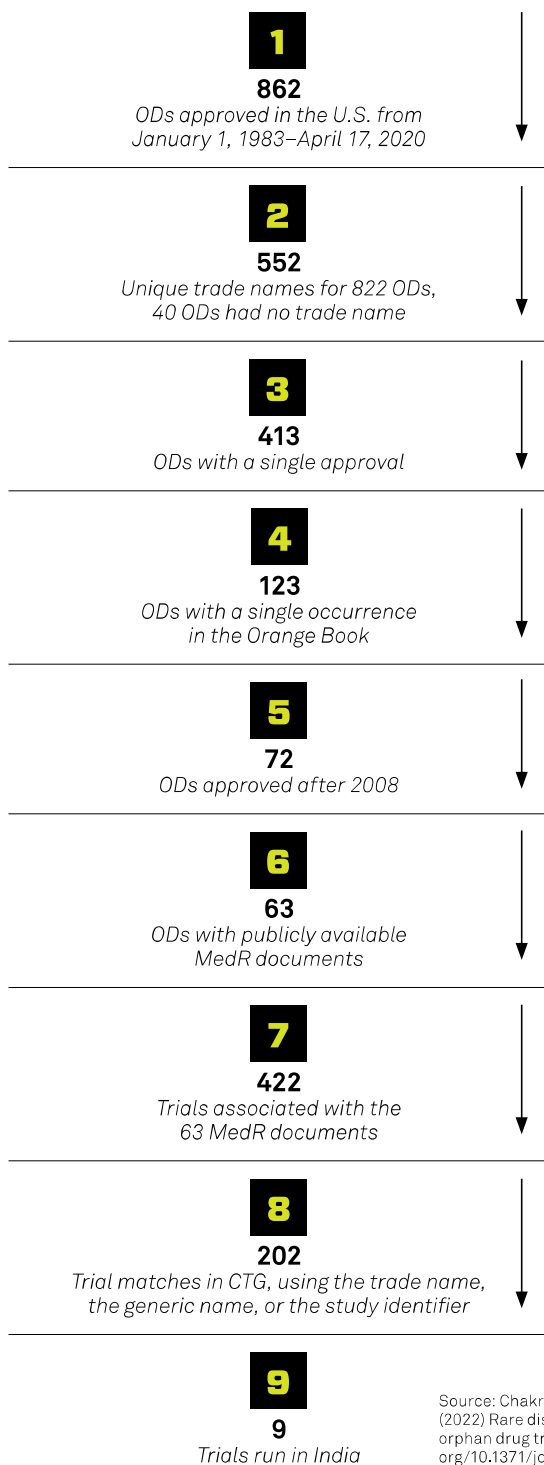
Additionally, the siloed nature of data capture and storage across different healthcare systems and the lack of standardized data-sharing practices hinder efforts to understand disease etiology, resulting in a skewed representation of patients in clinical trials with severe underrepresentation of patients from low- and middle-income countries like India. Despite being the most populous country in the world, accounting for almost a quarter of the global population representing a genetically diverse population, less than 1% of global rare disease clinical trials have study sites in India. As a result, orphan drugs are predominantly discovered and developed in the Western world, based on data from mainly Caucasian patients, reducing the generalizability of any treatment that may come to market.⁵

Harnessing Patient Registries for Research and Treatment

Patient registries have emerged as a crucial tool in addressing these challenges and accelerating rare disease research. Patient registries offer several benefits for advancing rare disease research and treatment. They are important tools to collect longitudinal data, facilitate collaborations, and promote knowledge sharing among researchers worldwide. They enable the identification and recruitment of eligible participants for clinical trials from across the globe, ensuring the representation of diverse patient populations, improving the generalizability of research findings, and helping to ensure that any treatment that emerges as a result of

Figure 1

The Steps Involved in Identifying the Orphan Drugs (ODs) and Trials of Interest



the research will benefit different patient populations.

Registries also facilitate the collection of patient-reported outcomes, helping researchers explore disease progression, identify unmet needs, and evaluate treatment effectiveness. By enabling long-term follow-up, registries make it possible to assess treatment outcomes, including measures of safety and quality of life in the long run.

Multistakeholder Collaborations to Power Patient Registries

Registries can be initiated and maintained by a wide variety of stakeholders, including patient foundations, biopharmaceutical sponsors, or academic investigators. It is essential that all stakeholders at the state and national levels of government play an important role in establishing and maintaining patient registries. There are a number of government-led initiatives that facilitate investment in infrastructure and technology to promote data sharing, interoperability, and the establishment of national and international rare disease networks. Governments also provide funding for rare disease research through dedicated research programs and grants and facilitate collaborations with regulatory bodies to streamline the drug approval process for orphan drugs.

In 2021, the Indian Council of Medical Research initiated a hospital-based National Registry for Rare Diseases to gather much-needed epidemiological data on rare diseases in the country.² Increased awareness and advocacy are needed for government stakeholders to recognize the unique challenges patients with rare diseases face. It is essential

Source: Chakraborty M, Choudhury MC, Chakraborty I, Saberwal G (2022) Rare disease patients in India are rarely involved in international orphan drug trials. PLOS Glob Public Health 2(8): e0000890. doi: [10.1371/journal.pgph.0000890](https://doi.org/10.1371/journal.pgph.0000890)

that governments and patient groups work together for the successful implementation of patient registries. Government can support registries through funding, regulatory guidance, and access to resources, and patient advocacy groups contribute through their advocacy and awareness initiatives and access to patient communities. Such joint initiatives can accelerate the development of patient registries, create standardized data collection protocols, and promote the sharing of research findings. Collaborations can also facilitate the establishment of patient-centered research networks, fostering multidisciplinary collaborations and enhancing the translation of research into clinical practice.

Collaborative effort is also necessary to overcome specific hurdles in rare disease research, such as poor identification of patient population due to the lack of effective diagnostic tools, unavailability of standardized protocols for data collection, and no consensus on data ownership, governance, and sharing. It is also essential to bring together stakeholders through international collaborations in order to engage the global rare disease community. Organizations such as the Indo US Organization for Rare Diseases (IndoUSrare) are working to foster such collaborations in an attempt to accelerate rare disease research.

Patient registries serve as a key tool in accelerating rare disease research, addressing obstacles such as limited data, siloed healthcare systems, and the need for representation of diverse patient populations. Patient registries offer opportunities for multistakeholder collaboration to unlock valuable insights into rare diseases. With support from the government and effective collaboration between governments and nonprofits, patient registries have the power to drive advancements in research and treatment, and ultimately improve the lives of patients worldwide. [GPI](#)

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