Protecting the Right to Health for Rare Disease Patients: Legal Frameworks, Policy Gaps, and the Need for Reform in India

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ABSTRACT
Right to Health is a fundamental human right recognized by various international and regional treaties, and governments worldwide have a duty to ensure its protection. However, several impediments exist for the full realisation of this right which includes economic, social and political factors. This requires coordinated efforts from governments, international organisations and civil societies.
Access to healthcare and medicines is central to this right. In the case of certain diseases, the full realisation of this is burdensome. This issue is reflected for individuals suffering from rare diseases, where orphan drugs, for their treatment are often unavailable due to the lack of incentives for pharmaceutical companies to invest in their development. Orphan diseases presents a complex dynamics due to the numerous diseases, varied and unidentifiable symptoms and difficulties in diagnosis.
Orphan drugs face significant hurdles in research, development, and commercialization, primarily due to the small market size and high costs involved. These barriers leave patients without access to life-saving treatments, thereby increasing health inequities. Several countries have introduced incentives for orphan drug development, such as market exclusivity, tax credits, research grants, fee waivers, accelerated approval etc. However, accessibility remains restricted due to high costs and insufficient governmental support.
India, with its large population suffering from rare diseases, lacks a robust regulatory framework to promote orphan drug development and ensure access for patients. Despite the introduction of the National Policy for Rare Diseases (NPRD) in 2021, there are significant gaps in incentivizing research and providing affordable treatment options. This paper examines the scope of right to health as a human right within both international and Indian contexts, analyzes the challenges associated with orphan drug development, and reviews the effectiveness of existing policies. It also explores global initiatives for development of orphan drugs, emphasizing the need for policy reform to ensure equitable healthcare for all.

INTRODUCTION

Health is a wide term corresponding to complete well-being which includes access and affordability of medicines and medical services; safe food and drinking water; nutrition, sanitation, etc. Right to health is an internationally recognized human right. Several international instruments, regional and human rights treaties call for the achievement of the best possible quality of health. Health is one of the top priorities of governments and is at the core of social welfare. It is the responsibility of states to protect this right and to shape their policies so that citizens fully enjoy it. Access to medical care is a global problem that equally affects developing and developed countries. Health emergencies and pandemics are a global concern and therefore a higher responsibility is placed on governments to prioritise health and ensure availability of medical facilities to all.
Medicines play an indispensable role in the prevention and treatment of diseases. Though there may be substitutes to address other components of treatment, nothing can completely substitute the role of medicines. Affordability and accessibility of medical facilities are important factors related to health. A substantial population of the world does not have regular access to essential medicines. Moreover, people with most serious health problems are the poor and underprivileged.
Millions of people around the world are denied basic healthcare and essential medicines due to economic and cultural barriers, with those suffering from rare or orphan diseases being one of the most vulnerable groups. As the name suggests, these diseases affect only a small number of people.
A medicinal product used in the treatment, prevention or diagnosis of a rare disease is known as an orphan drug. The development of such drugs presents significant hurdles that discourage manufacturers from investing in their research. Pharmaceutical companies prioritize profitability and tend to focus their resources on diseases with higher commercial potential. Bringing a drug to market requires substantial time and investment, not only

in the discovery phase but also in rigorous testing to ensure safety and efficacy. However, the small patient population and the complexities involved in developing orphan drugs make them less appealing to manufacturers, as the likelihood of recovering their investment remains low.

Lack of availability of medicines and inability to access drugs is detrimental to the patients suffering from orphan diseases. In aggregate this is not a small number. Since there is no medicine, they would be tied down to highly expensive treatments, which may or may not be fruitful. Hence, the society in a whole is at a loss since a significant amount of the population succumb to rare diseases every year.

Considering the challenges, various countries have enacted legislation providing incentives for development of orphan drugs. This has enhanced R&D in the field and many orphan drugs have been approved and marketed ever since. However, there are factors which could restrict access such as drug prices and government policies.

India is a country where a large population suffers from rare diseases. Lack of necessary drugs coupled with high treatment costs leads to mental and economic drain of the patients as well as their families. India also lacks a regulatory framework that adequately promotes research and development in orphan drugs and also addresses the patients' needs. The National Policy for Rare Diseases (NPRD) of 2021 was introduced with the aim of bringing down rare disease prevalence in India. It provides for measures of prevention and control and also deals with financial support. However, NPRD does not adequately address all the issues in relation to Orphan Drugs. Orphan drug development is not adequately incentivised and mechanisms for access are to be reviewed. Effective implementation of the policy is cumbersome due to constraints in economic resources.

Looking from the perspective of public health, it is important that there be enough medicines and these be available to those in need at affordable rates. Otherwise, it is a violation of the right to health. There cannot be any segregation of rare diseases and common diseases in this respect. Therefore, the needs of the patients of rare diseases are equally important and require state intervention. This paper examines the right to health as a human right within both international and Indian contexts. It explores the landscape of orphan drugs and orphan diseases, highlighting the scope and inherent challenges in drug development and access. It provides insights into initiatives by other nations to promote orphan drug development. It also reviews the Indian initiatives on orphan drugs, assessing their effectiveness and pointing out the need for reform.

HEALTH AS A HUMAN RIGHT

Health is fundamental to human well-being, serving as the foundation for both human dignity and social welfare. A healthy population can effectively contribute to economic and national development and social stability. Given its critical importance, health is enshrined as a human right in various international agreements. While recognising every individual's right to adequate standard of living, the Universal Declaration of Human Rights, covers health and medical care.[1] Nations vary in their economic capacities. Some may be capable of providing the best and advanced healthcare while others struggle to provide even primary care. The International Covenant on Economic, Social and Cultural Rights enshrines the enjoyment of 'highest attainable standard of health'. It places the obligation on states to take measures for full realisation of this right, which includes provisions for reduction of mortality, child development, hygiene, prevention, control and treatment of diseases and also for ensuring universal access to medical services.[2] The same has been reiterated by General Comment No: 14 by the Committee on Economic, Social and Cultural Rights. The United Nations Sustainable Development Goal number four deals with ensuring healthy lives and promoting well-being of all ages.[3] Enjoyment of highest standard of health has been reaffirmed by the resolution on the Right of everyone to the enjoyment of the highest attainable standard of physical and mental health in the implementation of the 2030 Agenda for Sustainable Development.[4]

The right to health, as a subset of economic and social rights, does not need to be fully realized immediately and is contingent upon each nation's resources and capabilities. However, states should make all possible efforts to provide the most basic and primary health care and must guarantee the right to the maximum possible extent

[1] Universal Declaration of Human Rights (adopted 10 December 1948) UNGA Res 217 A(III) (UDHR) art 25

[2] International Covenant on Economic, Social and Cultiral Right (adopted on 16 December 1966 entered into force on 3 January 1976) 993 UNTS 3 (ICESCR) art 12

[3] Transforming Our World: The 2030 Agenda for Sustainable Development (adopted on 25 September 2015) UNGA Res 70/1

[4] The right of everyone to the enjoyment of the highest attainable standard of physical and mental health in the implementation of the 2030 Agenda for Sustainable Development (adopted on 23 June 2017) UNGA Res 35/23

based on available resources.[5] Right to health encompasses availability and accessibility of good quality health care services without any discrimination. This signifies that there should be sufficient quantity of medicines and an assurance that they are safe and also physically and financially accessible. Moreover, States also have the responsibility to give due regard to specific needs of different groups. Furthermore, states also should take measures in the development of drugs and vaccines in areas where there is insufficient research or where research is underfunded.[6] This right has been embraced by nations through international commitments and also has incorporated it in their domestic legislation and policies.[7]

The TRIPS agreement[8] while laying down minimum standards for the protection and enforcement of intellectual property rights, honours the right to health. The DOHA declaration[9] assures that implementation of TRIPS shall be in consonance with member states' efforts to protect public health. Moreover, the flexibilities provided in the TRIPS agreement aim to promote access to medicines and health technologies.
Under the Indian Constitution, right to health is inherent in the Right to life and personal liberty.[10] Moreover, the state is responsible for improvement of public health and standard of living.[11] The National Health Policy covers all pillars of health including access to good quality services at affordable rates and reducing health disparities. One of the fundamental principles of the National Health Policy is Universal Health Coverage aimed to be achieved through various programmes and schemes. The Supreme Court in various rulings has held the right to health as part of Right of life.[12] The duty of the state for providing adequate medical aid as part of right to life under Article 21 was emphasised by the Supreme Court in *Paschim Banga Khet Mazdoor Samity v. State of West Bengal*[13] and *Vincent Panikurlangara Vs. Union of India*.[14] The relation between right to health and the duty of states has been highlighted in the case of *State of Punjab and Ors. Vs. Ram Lubhaya Bagga*.[15]
Access, availability and quality are therefore inherent elements of right to health which is basic to human dignity and welfare. States must honour their obligations by making concerted efforts to advance health care equity, promote research and development, and ensure that all individuals can access essential health services.

[5] United Nations Human Rights Office of the High Commissioner, 'Fact Sheet No. 21: The Right to Health' (1 June 2008) <https://www.ohchr.org/sites/default/files/Documents/Publications/Factsheet31.pdf> accessed 5th September 2024.
[6] ibid
[7] International Convention on the Elimination of All Forms of Racial Discrimination (adopted 21 December 1965, entered into force 4 January 1969) UNGA Res 2106 (XX) ICERD art 5; Convention on the Elimination of All Forms of Discrimination against Women (adopted on 18 December 1979, entered into force 3 September 1981) 1249 UNTS 13 (CEDAW) art 11, 12, 14; Convention on the Rights of the Child (adopted 20 November 1980, entered into force 2 September 1990) 1577 UNTS 3 (UNCRC) art 24; Convention for the Protection of Human Rights and Fundamental Freedoms (European Convention on Human Rights, as amended) (ECHR) art 3 ; African Charter on Human and Peoples' Rights (adopted 27 June 1981, entered into force 21 October 1986) (1982) 21 ILM 58 (African Charter); Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights (Protocol of San Salvador) (entered into force 16 November 1999) OAS Treaty Series No 69 (1988) reprinted in Basic Documents Pertaining to Human Rights in the InterAmerican System OEA/Ser L V/II.82 Doc 6 Rev 1 at 67 (1992)
[8] General Agreement on Trade-Related Aspects of Intellectual Property (15 April 1994) 1869 U.N.T.S. 299 https://www.wto.org/english/docs_e/legal_e/27-trips.pdf
[9] Declaration on the TRIPS Agreement and Public Health, Doha WTO Ministerial Declaration 14 November 2001) https://www.wto.org/english/res_e/booksp_e/ddec_e.pdf
[10] The Constitution of India art. 21
[11] ibid art 47
[12] Bandhua Mukti Morcha v. Union of India, (1984) 3 SCC 161; Consumer Education and Research center and Ors. Vs. Union of India, (1995) s SCC 42; Consumer Education and Research center and Ors. vs. Union of India, (1995) 3 SCC 42
[13] (1996) 4 SCC 37
[14] (1987) 2 SCC 165
[15] (1998) 4 SCC 117

ORPHAN DRUGS AND ORPHAN DISEASES

In common terms we may understand orphan disease or rare diseases as those affecting a small number of people, in comparison to common diseases. Orphan drugs are used in the prevention or treatment of rare diseases. However, this is a dynamic and complex landscape.

There is no uniform definition for the term rare disease.[16] According to the WHO, a disease with prevalence of one or less per thousand population is deemed to be a rare disease.[17] Countries have adopted definitions suitable for their population and healthcare system. In the United States, orphan disease is a disease that affects less than two hundred thousand people.[18] As per the European Union Regulations, rare disease means a disease where the number of people affected is less than five per ten thousand people.[19] Australia has a similar definition. In Japan, diseases affecting less than fifty thousand people are termed as rare diseases.[20]

Almost six thousand to eight thousand rare diseases have been identified. In aggregate, a substantial population is affected by orphan diseases.[21] Sickle cell anaemia, Gaucher's disease, Cystic Fibrosis, Haemophilia, Thalassemia, Fabry disease are examples. Many diseases are genetic; others may be due to environmental factors, immune responses, etc. The prevalence is also varied among populations and countries. Some would be rare only in certain countries. They may be acute or chronic. They have varied symptoms and may be seen in people of all ages. Some are preventable and others are not. Treatment exists for some diseases and those may be expensive. Accurate and timely responses are also challenging.[22] Lack of clinical alternatives, difficulties in diagnosis, and lack of knowledge of health professionals, results in a heavy burden on the patient and their family.[23]

Development of orphan drugs to treat or prevent rare diseases also presents a complex picture. Due to varied and non-identifiable symptoms of rare diseases, discovering a chemical entity that pertains to a clinical

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[16] ORPHAN DRUGS AND RARE DISEASES, (David C. Pryde & Michael J. Palmer eds., 2014); RARE DISEASES AND ORPHAN PRODUCTS: ACCELERATING RESEARCH AND DEVELOPMENT, (Marilyn J. Field et al. eds., 2010); Bao Cheng Liu et al., *A Cross-National Comparative Study of Orphan Drug Policies in the United States, the European Union, and Japan: Towards a Made-in-China Orphan Drug Policy*, 31 JOURNAL OF PUBLIC HEALTH POLICY 407 (2010); Todd Gammie, Christine Y. Lu & Zaheer Ud-Din Babar, *Access to Orphan Drugs: A Comprehensive Review of Legislations, Regulations and Policies in 35 Countries*, 10 PLOS ONE 1 (2015); LILI LOORAND-STIVER, TARA COWLING & CHRISTINE PERRAS, *Drugs for Rare Diseases: Evolving Trends in Regulatory and Health Technology Assessment Perspectives*, (2013); Proteesh Rana & Shalini Chawla, *Orphan Drugs: Trends and Issues in Drug Development*, 29 JOURNAL OF BASIC AND CLINICAL PHYSIOLOGY AND PHARMACOLOGY 437 (2018).

[17] Michael Abramowicz, *Orphan Business Models: Toward a New Form of Intellectual Property*, 124 HARVARD LAW REVIEW 1362 (2011); Pedro Franco, *Orphan Drugs: The Regulatory Environment*, 18 DRUG DISCOVERY TODAY 163 (2013); Viswanath Pingali & Neelima Das, *Rare Diseases Require Support Too*, 46 VIKALPA 129 (2021).

[18] Gammie, Lu, and Ud-Din Babar, *supra* note 16; Liu et al., *supra* note 16; Franco, *supra* note 17.

[19] Mari Minn, *Development of Orphan Drugs under European Regulatory Incentives and Patent Protection*, 24 EUR. J. HEALTH L. 239 (2017); Gammie, Lu, and Ud-Din Babar, *supra* note 16; Liu et al., *supra* note 16.

[20] Gammie, Lu, and Ud-Din Babar, *supra* note 16; Liu et al., *supra* note 16; Franco, *supra* note 17.

[21] RARE DISEASES AND ORPHAN PRODUCTS, *supra* note 16; Helen Crompton, *Rare Diseases and Orphan Medicinal Products*, MANCHESTER METOPOLITAN UNIVERSITY RESEARCH REPOSITORY, https://e-space.mmu.ac.uk/1638/1/crompton%20wp02_03.pdf; Xaviere Giroud, *Dancing with Our Hands Tied: An Imbalanced Focus on Drugs for Orphan Disease and Research*, 16 NORTHWESTERN JOURNAL OF TECHNOLOGY AND INTELLECTUAL PROEPRTY 285 (2019); LOORAND-STIVER, COWLING, AND PERRAS, *supra* note 16.

[22] RARE DISEASES AND ORPHAN PRODUCTS, *supra* note 16; LOORAND-STIVER, COWLING, AND PERRAS, *supra* note 16.

[23] R. Rodriguez-Monguio, T. Spargo & E. Seoane-Vazquez, *Ethical Imperatives of Timely Access to Orphan Drugs: Is Possible to Reconcile Economic Incentives and Patients' Health Needs?*, 12 ORPHANET J RARE DIS 1 (2017).

indication would be difficult. Since the affected population is smaller, conduct of clinical trials to prove the safety and efficacy of a drug would be difficult.[24]

In case of orphan drugs, the natural history and pathophysiology may not be completely known therefore making it difficult to identify drug targets and the relevant study endpoints and biomarkers. Preclinical studies identify the pharmacokinetic properties and their toxicity. Sufficient animal models become important here in proving efficiency of selected drugs and they might not be available in the case of orphan drugs. During the clinical trial stage, developers face various challenges such as achieving adequate sample size. It is cumbersome to identify patients and bring them together. However, once patients are identified, they are easier to retain and the dropout rate is also less. Further, patients with rare diseases are more likely to accept new medication or experimental treatment.[25] Therefore, research and development in orphan drugs involves high costs and resources.

Orphan drugs may be divided into two categories – new molecular entities developed for a rare disease and repurposed drugs that are already approved for other diseases and repurposed for a rare disease. For development of a new molecular entity, the process begins with understanding the pathology of a disease, then target selection and clinical trials to prove the safety and efficacy of the drug. [26]

Drug manufacturers would be interested in recouping their investments through sales. Hence their research would be directed to those drugs with high demand since they guarantee huge returns. The small market size makes orphan drugs not commercially viable and hence does not attract manufacturer interest since the returns will not compensate the high costs involved in research and development.

Intellectual property rights also have an important role in the case of pharmaceuticals. Patent protection have helped the pharmaceutical industry grow and flourish since the exclusivity ensures recovery of the research expenses. The exclusive right is granted in exchange of the disclosure of the invention to the public. This fosters further research and the ultimately the society is benefitted with new and more efficient products. Moreover, patent also safeguards manufacturers from genetic competitors. It is common knowledge that in order to be patentable the invention has to be novel, non-obvious, and has to have industrial utility.

However, these conditions could also act as barriers to drug development especially in the context of orphan drugs. Orphan drugs need not necessarily possess the 'novel' and 'non obvious' aspect since most of them might be enhancement of the properties of already known products. The field of orphan drugs itself being commercially discouraging, absence of exclusivity will again make researchers abandon their projects or not to take up projects at all. Manufacturers would also filter drugs based on their eligibility for intellectual property protection to ensure exclusivity. This acts as a barrier to free-riding and generic competition.[27] Hence they are reluctant to take up research where they do not get complete exclusivity (expired patents or repurposing of drugs) and hence those drugs that may be socially relevant and can cure serious health crises will never be developed.[28] Moreover, exclusivity has its often effects as a barrier to access.

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[24] ORPHAN DRUGS AND RARE DISEASES, *supra* note 16; THERESA M. WIZEMANN, SALLY ROBINSON & ROBERT B. GIFFIN, BREAKTHROUGH BUSINESS MODELS: DRUG DEVELOPMENT FOR RARE AND NEGLECTED DISEASES AND INDIVIDUALIZED THERAPIES: WORKSHOP SUMMARY (2009); RARE DISEASES AND ORPHAN PRODUCTS, *supra* note 16.

[25] ORPHAN DRUGS AND RARE DISEASES, *supra* note 16; Rana and Chawla, *supra* note 16.

[26] Rana and Chawla, *supra* note 16.

[27] Christopher Buccafusco & Jonathan S. Masur, *Drugs, Patents, and Well-Being*, 98 WASH. UL REV. 1403 (2021); Frederick M Abbott, *Health and Intellectual Property Rights*, *in* RESEARCH HANDBOOK ON GLOBAL HEALTH LAW 135 (2018); German Velasquez, *The Right to Health and Medicines: The Case of Recent Multilateral Negotiations on Public Health, Innovation and Intellectual Property*, 14 DEVELOPING WORLD BIOETHICS 67 (2014).

[28] ORPHAN DRUGS AND RARE DISEASES, *supra* note 16; RARE DISEASES AND ORPHAN PRODUCTS, *supra* note 16; WIZEMANN, ROBINSON, AND GIFFIN, *supra* note 24; ELIZABETH HERNBERG-STÅHL & MIROSLAV RELJANOVIĆ, ORPHAN DRUGS: UNDERSTANDING THE RARE DISEASE MARKET AND ITS DYNAMICS (2013); Rana and Chawla, *supra* note 16.

LEGISLATIVE INCENTIVES TO PROMOTE R&D IN ORPHAN DRUGS: A GLOBAL PERSPECTIVE

Rare diseases therefore remained an ignored field not receiving attention from manufacturers. However, the promotion of research and development was imperative since the patient pool is very high. This necessitated regulatory intervention since the current system did not properly incentivise orphan drug development. Public health demands motivated various nations to intervene and introduce economic incentives for the development of orphan drugs.

The United States was the first to introduce legislation in this respect.[29] This inspired various other nations to introduce legislation or make amendments to their existing legislation. In the European Union, Regulation 141/2000 was implemented to deal with orphan drugs. In Australia, necessary amendments were made to the Therapeutic Goods Act, 1990 in order to incorporate orphan drugs. In Japan, the Pharmaceutical Affairs Law was revisited to encourage more research and development in Orphan Diseases. Legislations exist in various other countries as well.[30]

Market exclusivity is the primary incentive granted although the duration differs according to countries. If a drug is designated as orphan according to national standards, then it will be granted a period of exclusivity within which a similar product will not be granted market approval unless the second applicant is able to demonstrate clinical superiority. In Japan, a re examination period of 10 years is provided under the Japanese Law during which other similar applications cannot be submitted for market authorisation. Australian law does not provide for market exclusivity. Other incentives include tax credits, scientific assistance, fast tract application, exemptions, and waivers in fees.[31]

The regulatory incentives promote orphan drug development since the industry will be unwilling to develop orphan drugs under normal market conditions and will not be able to recoup their investment through sales. However, orphan drugs ought to show the same standards of safety and efficiency as any other drug.[32]

Introduction of incentives have radically changed the orphan drug landscape leading to increased number of approvals and market authorisations. For instance, in the United States, more than six thousand designations have been granted for various clinical indications till date and more there are more than thousand approvals.[33] Gradually, a good commercial opportunity has also been seen in the market since the prices of orphan drugs are high. Studies indicate that the most expensive drugs are orphan drugs.[34] Moreover, drug manufacturers would employ various strategies to sustain their exclusivity.

However, high prices of drugs limit patient access to orphan drugs. From the regulatory angle, a policy must focus not only on the development of orphan drugs but also that patients get timely access to safe and efficient drugs. Regulatory incentives although have resulted in increased R&D in orphan drugs; the incentives do not holistically address the needs of the patient; they neither provide for long-term innovation nor solve the problem of affordability. Therefore, there is a need to align incentives to companies and societal concerns.[35]

ORPHAN DRUGS LANDSCAPE IN INDIA: NEED FOR REFORM

Rare diseases and Orphan drugs have only seldom popped in policy formulation. Despite India being one of the countries with the most prevalence of rare diseases and most of the patient being from the poor and under privileged, no comprehensive measure for prevention and cure of these have been taken till now. As per the New Drugs and Clinical Trial Rules, 2019[36] orphan drugs mean medicines used to treat a condition which

[29] Orphan Drug Act of 1983, Pub. L. No. 97-414. 96 Stat. 2050 (1983).

[30] Gammie, Lu, and Ud-Din Babar, *supra* note 16; Liu et al., *supra* note 16.

[31] Bao-cheng Liu et al., *A Cross-National Comparative Study of Orphan Drug Policies in the United States, the European Union, and Japan: Towards a Made-in-China Orphan Drug Policy*, 31 JOURNAL OF PUBLIC HEALTH POLICY 407 (2010); Gammie, Lu, and Ud-Din Babar, *supra* note 16.

[32] Laetitia Benard, Jacqueline Bore & Eveline Van Keymeulen, *Has the Orphan Regulation Met Its Aims*, 2 EPLR 179 (2018).

[33] Lewis J. Fermaglich & Kathleen L. Miller, *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*, 18 ORPHANET J RARE DIS 163 (2023).

[34] Rana and Chawla, *supra* note 16; LOORAND-STIVER, COWLING, AND PERRAS, *supra* note 16.

[35] Rodriguez-Monguio, Spargo, and Seoane-Vazquez, *supra* note 23.

[36] The New Drugs and Clinical Trial Rules, 2019, Gazette of India, pt. II sec. 3(i) (February 1 2018).

affects less than five lakh people in India. The Rules further provide for relaxation of conditions for clinical trials in the case of rare diseases or orphan drugs. It also provides for speedy approval process and fee relaxations.

National Policy for Rare Diseases

The Delhi High Court in *Mohd. Ahmed (Minor) v. Union Of India & Ors*[37] stressed on the need for a national policy on rare drugs and formulation of committees for the same. Based on the recommendation of the V K Paul Committee, I C Verma Sub Committee and D K Tempe Interdisciplinary Committee, The National Policy for Treatment of Rare diseases 2017 was formulated. This was withdrawn due to the lacunas and also due to challenges in implementation. The National Policy on Rare Diseases, 2021 revamped the earlier policy and attempted to bring a more sophisticated approach to rare diseases.[38]

The policy notes the varying definitions of rare diseases and points out indicators which are relevant in formulation of a definition of rare disease. Other than disease prevalence, factors such as severity, location, level of rarity etc. also contribute in forming the definition of rare disease. Lack of primary care professionals and facilities are hurdles in diagnosing rare diseases since many a times traditional genetic testing might not point out the exact problem. Lack of awareness and training might lead to wrong diagnosis which may severely threaten the patient's life. Lack of treatment and high costs just increases the burden of the patients and their families.

The policy notes that there is a lack of data which would enable analyses of the prevalence of rare diseases to enable formulation of a definition specific to the Indian context. A National Registry is being initiated under the policy. The policy categorises the known rare diseases into different groups based on the available knowledge. Prevention and control of rare diseases aims to be achieved and Centres of Excellence are established for enhancing the capacity of health professionals and for diagnosis and treatment of rare diseases. The policy also provides for providing financial support to patients and also for crowdfunding. The policy also envisages promotion of Research on rare diseases and development of drugs by involving funding agencies. An integrated approach involving various ministries is sought for encouraging local manufacture of orphan drugs. The Department for Promotion of Industry and Internal Trade has been requested to promote local manufacture of drugs.

Almost four hundred and fifty rare diseases have been identified in India. Although precise numbers are not there, almost ninety million people in India are affected by rare diseases by adopting international estimates. This has been acknowledged in the 2017 policy. The Indian Council of Medical Research launched the National registry for rare diseases in 2019. Till now about fifteen thousand cases have been registered.[39] The National Consortium for Research and Development on Therapeutics for Rare Diseases in India was established in 2021 to promote research, development and technology transfer of therapeutics for orphan drugs.[40] There are fifty-seven ongoing projects on rare diseases receiving funds from various agencies.

The NPRD though is a positive step, there is still a long way to go. The policy does not adequately incentivize orphan drug development so that research and development happen at a large scale bypassing all the challenges posed by orphan drugs. A national registry can ensure collection of data and improvement from the health perspective. Government support is also essential for funding the institutions researching this sector. Lack of awareness and social stigma are also hurdles on the way. Moreover, its strict implementation and enforcement would be problematic. Therefore, those in need are again put to agony.

[37] 2014 SCC OnLine Del 1508.

[38] National Policy for Treatment of Rare Diseases, 2021 available at: https://rarediseases.mohfw.gov.in/uploads/Content/1624967837_Final-NPRD-2021.pdf;

[39] Department of Health Research Ministry of Health and Family Welfare Government of India, Rare Diseases Registry available at: https://rdrdb.icmr.org.in/registry/

[40] Department of Health Research Ministry of Health and Family Welfare Government of India, Establishment of National Consortium of Research and Development on Therapeutic for Rare Diseases, https://rdrdb.icmr.org.in/static/Establishment.pdf

Way forward

India is therefore in need of a legislative enactment that balances both the developer's needs as well as that of the patients. Firstly, it becomes essential for forming a definition of orphan drugs that is suited to the Indian scenario. It has to be flexible and has to have a wider ambit. This ensures that the cure for various diseases could be covered, thus remedying the lacunas in the existing system so that ultimately the society is benefitted with more and efficient medicines. The definition should incorporate disease prevalence as well as other connected factors such as disease severity, level of rarity, causes etc. India also needs a clear and distinct process of designation and market approval under a separate authority so that there is no unwarranted delay.

Regulatory incentives should also be introduced to encourage research in orphan drugs. This can promote domestic manufacture and therefore keep prices under control. However, provision for market exclusivity should be modified according to Indian demography. In addition to protection, availability of sufficient funds is also important. Due to the distinct nature of orphan diseases, institutions/organisations/agencies would not be ready to fund research activities as they cannot expect sufficient returns. Moreover, as provided in the NPRD, government institutions such as CSIR should actively take up research in orphan drug development.

Funds are necessary not only for supporting patients for treatment but also for drug development. Due to economic constraints of the government diversion of adequate funds for orphan drug development remains a pertinent question. Hence new sources ought to be found and the legislative policy should envisage funding from these other sources. The strategy must also acknowledge ethical considerations and prioritize patient needs. Public – Private partnerships, technology transfer and data sharing should also be important considerations of the policy.

In addition to protection, accessibility is also a significant concern. For patented products mechanisms such as compulsory licensing and parallel importation enables the state to ensure availability of orphan drugs. The potential of similar schemes can be explored for orphan drugs also. It is equally important that drugs reach those in need, at affordable rates. Drug pricing ought to be effectively regulated. These may be provided under government schemes. Drug prices may be subsidized by utilizing CSR initiatives of businesses. Bringing such a mechanism would ensures the health and well-being of everyone leaving no exception. Programs under the National Health Mission such as RMNCH+A can be useful in the management of rare diseases such as adopting preventive strategies, in early detection and diagnosis. The scope of the treatment under various programs of NHM can be broadened to include rare diseases.[41]

CONCLUSION

Right to health is one of the top most priority of the nation and therefore, health initiatives should have a universal ambit covering various dimensions. The orphan drug concept forces society to consider whether the right to health is truly universal, or if economic and market considerations compromise its reach.

Orphan drugs and orphan diseases therefore requires more attention from the government and needs to be addressed as that of any mainstream disease. The scope of right to health places obligation on states to take proactive measures for promotion of research and development in underfunded and neglected areas. The principles of universal health care and non-discrimination emphasise that the needs of patients with rare diseases be no longer considered secondary. Absence of availability of orphan drugs and lack of access to drugs stands in direct violation of their right to health.[42]

Many nations have brought in policies that provide incentives for orphan drug development which has contributed to increased R&D. However, there is lack of uniformity and international consensus. Though the policies vary in their definition and implementation, the introduction of a legislative framework have significantly changed the R&D landscape. Despite the increased number of approvals, the needs of all patients are not met. The incentives have become too generous that they have been exploited. Thus, indicating the health inequities and the need for optimal allocation of resources. Considering the increasing number of rare diseases and the increased expenses in treatments, clear ethical guidelines have to be brought.

India is a country where the Orphan drug landscape is largely ignored. There is a huge population in India especially among underprivileged sections who need affordable medicines for rare diseases. Since there are no substantive steps towards a binding enactment, the demands remain unmet. Hence India is in desperate need of a legislative enactment.

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[41] Mohua Chakraborty Choudhury & Pragya Chaube, *Integrating Rare Disease Management in Public Health Programs in India: Exploring the Potential of National Health Mission*, 17 ORPHANET J RARE DIS 43 (2022).
[42] Crompton, *supra* note 21; Rana and Chawla, *supra* note 16.

The new approach for the protection of orphan drugs and their domestic manufacture ought to be adopted. Also, instead of a single payment provided to one category of rare diseases, routine payments based on the severity of the disease could be provided by the government by utilising funds from other sources. Collection and consolidation of data is an important step here, so that patient may be registered under the government for enjoying the fruits of the government schemes

Orphan drug development ought to be promoted keeping in mind the principles of affordability and accessibility. Being a developing country, the aim is not to bring more expensive treatment and high-cost medicines; but ensure that all those in need get adequate care and treatment. The new approach should also address the unique challenges posed by orphan drugs such as flexible clinical trials guidelines and small market size.

Addressing the challenges relating to orphan drugs require a careful balance between encouraging innovation and ensuring that the fruits of that innovation are available to all. Drug development ought to be encouraged for rare diseases but at the same time, value-based pricing should also be given importance. It is necessary for approving drugs which may satisfy the patients' needs. By rethinking policies related to orphan drugs and promoting global cooperation, we can move closer to realizing the promise of health as a fundamental human right for everyone, including those with rare diseases.

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