A Review on National Policy of Rare Diseases, 2021

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Abstract

In a country like India that already struggles to provide basic health facilities to its vast population, it becomes very difficult for people suffering from rare diseases to access treatment options. Currently, about 96 million people in India are living with a rare disease with almost no access to any kind of treatment. The National Policy of Rare Diseases was approved in 2017 but had limited impact as it faced a lot of implementation challenges. In the light of this, the policy was reframed after stakeholder's consultation and reviews from an expert committee. The reframed 'National Policy of Rare Diseases' was approved in March 2021. The study attempts to critically evaluate the policy in terms of its reach to the wider population and access to treatment for the most vulnerable population in India. An in-depth analysis reveals several limitations of the newly designed policy. The lack of availability of data to define the disease as per Indian standards, the absence of a framework for communication between the stakeholder institutions at all levels, and the lack of incentive to domestic drug manufacturers to invest funds into the research and development of the medicines for rare diseases severely limits our ability to deal with this public health concern. The Government needs to work towards developing a more holistic and empathetic policy to address the issues of lakhs of people suffering from rare diseases in India.

INTRODUCTION

Rare diseases are diseases that affect a very small percentage of the overall population. Globally, there is no unanimity on a cut-off number for a disease to be considered rare. WHO does have its definition, but different countries have different definitions based on studies conducted in their country and the rarity of the disease. The United States defines a rare

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disease as a disease that affects less than 20,000 people in the country. (Genetic and Rare Diseases Information Center (GARD)Europe, Canada, Japan, South Korea, and Australia consider a disease as a rare disease if the prevalence of the disease is less than 5.0, 5.0, 4.0, 4.0,1.0 per 10,000 population, respectively. ("National Policy for Rare Disease, 2021) A study states that the average prevalence thresholds used to define rare diseases ranged among different jurisdictions from 5 to 76 cases/100,000 people, with a global average prevalence threshold of 40 cases/ 100,000 people (Richter et al. 2015) This range of cases varies based on different factors such as location, level of rarity, study ability, etc. There is an exhaustive list of rare diseases which list 7000 diseases considered as rare diseases.

These rare diseases are majorly thought to be genetic and are passed on from one generation to the next. There are many rare diseases like autoimmune diseases and cancers, that might not be inherited ("FAQs about Rare Diseases | Genetic and Rare Diseases Information Center (GARD) – an NCATS Program" 2017). Rare diseases are also known as orphan diseases since the market is not large enough to invest money and resources into research and development for discovering treatments of such diseases. Progress is being made on different fronts in terms of prevention, diagnosis, and treatment. Different countries have policies in place to deal with rare diseases affecting their population.

RARE DISEASES AS A PUBLIC HEALTH CHALLENGE IN INDIA

To address challenges pertaining to rare diseases requires collection and study of complex epidemiological data, challenges in research and development for prevention and treatment, accurate diagnosis, and a robust tertiary healthcare system that involves long-term care, including the high costs of treatment. With the limited availability of resources, it becomes a tough call for India to decide between increasing spending on general healthcare problems, which will benefit a large number of populations in compare to allocating more significant resources towards a relatively small number of people suffering from these rare diseases.

In the majority of the cases, these rare diseases are serious, debilitating, chronic, and life-threatening. The treatments are specialized and long-term. Children are disproportionately affected by these diseases as compared to adults. 50 percent of new cases are observed to be in children, out of which 35 percent of children die before the age of one year, 10 percent die between the ages of 1 to 5 years, and 12 percent between the ages of 5 to 15 years. (Ministry of Health and Family Welfare 2017) The trauma on families of patients suffering from rare diseases is emotional as well as financial.

India does not have its standard definition for rare disease and neither does sufficient data on prevalence exists. The Government of India launched the Indian Rare Disease Registry only in April 2017. Only 450 rare diseases have been recorded in the registry as per data available from tertiary hospitals. Although no official data exists on the number of people in India, suffering from rare diseases, an estimated 70 million people in India are victims of a rare disease (Kurian, Krishnan, and Sappani 2021). Approximately 50 percent of the rare diseases are onset at birth. The most common reported rare diseases are Thalassemia, Haemophilia, Sickle-cell Anaemia, auto-immune diseases, Primary Immuno Deficiency in children, Hirschsprung disease, Gaucher's disease, Lysosomal storage disorders such as Pompe disease, Cystic Fibrosis, Hemangiomas and certain forms of muscular dystrophies.

POLICY INITIATIVES AND CHALLENGES IN THE CONTEXT OF INDIA

The National Policy on Rare Diseases was first put out by the Central Government in 2017. The policy was put on hold citing implementation challenges. As pointed out by the States, some issues with the policy were lack of clarity regarding cost-sharing, disease coverage, and patient eligibility for getting treatment of the rare disease under this policy. To address these queries, a review committee was formed in 2018 which submitted its recommendations in January 2021, following which further consultations were held. The new National Policy on Rare Diseases was finally released in March 2021(Perappadan and Koshy 2021). Despite addressing the challenges of the previous policy by providing guideline under which scheme of central government and other clarification, the new policy still falls short of developing a strong framework, with processes that might make treatment affordable and accessible for the commoner.

India does not have its definition of a rare disease. The new Policy states that sufficient data is required to arrive at a definition that will be best suited to India. A National Registry was set up by ICMR to prepare a database of rare diseases in India in 2017 for the same. Under this initiative, the Government identified centres and institutions that have the facility of qualified investigators for the identification and diagnosis of rare diseases. Only 12 States and 4 Union Territories in the entire country have approved institutions with such facilities available, with even bigger states like Gujarat, Madhya Pradesh, Kerala, etc being left out ("Centers" 2017). As a result, the registry has not made any significant progress in terms of data collection, with only 450 diseases being available on record as of now. There cannot be any informed decision-making without the availability of data. With a lack of sufficient data, it takes up to seven years for a patient

to be diagnosed with a rare disease. It takes a minimum of three misdiagnosis before a correct assessment is made for a patient and treatment is started (Economic Times 2021b) the diagnosis becomes almost difficult without the database, and precious time is lost in starting the correct treatment.

The policy suggests notifying nine institutes as Centre of Excellence which will act as premier tertiary hospitals with facilities for prevention, treatment, and diagnosis of rare diseases. Similarly, the setting up of five Nidan Kendras is suggested for screening, testing, and counselling of rare diseases and also provide treatment if the facilities exist. For pan India coverage, the policy heavily relies on the health care workers to undertake screening activities for the early diagnosis of diseases. It becomes very evident that there is a need to layout a detailed procedure, which underlines the steps to be taken after the disease is diagnosed by a health care worker. The patient needs to be guided in terms of whom to approach at every level of diagnosis, referral to institutions, and treatment. There also has to be a framework for reporting the screening and diagnosis data from health care workers to tertiary hospitals, Centres of Excellence, and Nidan Kendras. The policy does not lay out clear guidelines regarding the coordination required among the various levels between all stakeholder institutions. A country like India with a significantly higher chunk of masses must have the appropriate service delivery framework. In which, policymakers should bifurcate the responsibilities at the block and district level. One nodal institute at state will not be sufficient to detect and prevention of rare genetic diseases. The government of India has tackled various major health problems such as leprosy, dengue, tuberculosis, Chikungunya, malaria etc., by ensuring adequate service delivery. The policy has not defined any such mechanism to address such service delivery issues. Counselling and guidance centres should be in place at least at the district level to guide people suffering from rare disease in terms of prevention and cure.

Moreover, there is no referral mechanism to access the NidanKendras and centre of excellence stated in the policy.This would result in the unavailability of data to build the registry. Furthermore, this lack of framework would make the treatment inaccessible to the commoner. In the previous policy there was no clarity on cost sharing, the new policy has addressed this challenge and states that cost will be taken care by central government but again there in no clarification on implementation of the proposed benefits. Cross controlling mechanism for diagnosis prevention and disbursement of financial assistance is lacking in the policy.

Under the policy, central government will financial assistance of up to Rs 20 lakh to each individual suffering from a rare disease as one-time assistance for treatment under the umbrella scheme of Rashtriya Arogya Nidhi. Diagnosis is a challenge in the case of rare diseases. Even after a diagnosis is made, no treatment is available for a majority of rare diseases. Available research suggests that despite the recent advancements in this field, only 5 percent of the diseases have a cure (The Lancet Diabetes & Endocrinology 2019). In the case of certain rare diseases, even though no cure exists, the person can live a longer and improved life with medicines. The budget of the government towards public health expenditure is limited and with resource constraints. For effective utilization of the limited budget cases in which the diseases have an existing cure should be prioritized to save maximum lives. Current policy states that it will provide the same financial assistance of 20 lakh to all diseases whether it costs ten lakh or ten crores. Disease wise financial assistance cap needs to be defined for effective utilization of funds, and a more concentric approach to the problem is required.

Moreover, there is no guideline on empanelment of hospital and claim of funds in case of any rare disease diagnosed. The policy has stated that Separate arrangements should be made for cases with no cure and requiring lifetime support of medicines and pain management therapies under the policy.

It is stated in the policy that pharmaceutical industries would be encouraged to develop drugs for rare diseases. The Department of Pharmaceuticals, Department for Promotion of Industry and Internal Trade (DPIIT) will be roped in to create a conducive policy framework for domestic pharmaceutical companies to focus on the research development and manufacturing of indigenous drugs for curing rare diseases with the provision of tax benefit and research grants along with notification of developed drugs as notified drugs which will be available at affordable rates. Currently, the number of pharmaceutical companies manufacturing drugs for rare diseases is very few. There is no domestic manufacturer in India except for the Food for Special Medical Purposes (FSMP) that manufactures food formulations for nutritional support to patients suffering from certain medical conditions. However, too much focus on making the drugs affordable when there is a complete absence of interest from the companies may not be such a good idea.

For example, to solve this problem, a pioneering decision was taken by the United States Government in 1982 by passing the Orphan Drug Act. Before its passage, only ten drugs existed for orphan diseases; by today, that number had grown up to more than 600, which highlights the Act's

success (Huron 2021). The Act's incentives, coupled with the fact that pharmaceutical companies can set any price they like for a drug in the US, have made the orphan drug market astonishingly profitable. An analysis found that the 'economics and investment case for orphan drug development' was 'more favorable than for non-orphan drugs' (Meekings, Williams, and Arrowsmith 2012). Pharmaceutical companies, thus, generate greater returns by developing orphan drugs rather than developing drugs for common diseases.

After the act was passed in 2010, about 30 percent of drugs approved by the FDA were orphan drugs, even though only 10 percent of the population is affected by orphan diseases. Moreover, out of the total drug sales in 2012, more than 30 percent of sales were accounted for by orphan drugs. The market for orphan drugs is expected to grow at double the rate of the normal prescription drug market by 2022(Delton n.d.).

At a conference held by the Indian Drugs Manufacturers Association in 2001, a group of pharmacologists requested the Indian Government to formulate the Orphan Drug Act in India (Sharma et al. 2010). A lack of interest from concerned authorities, combined with the absence of any legislation aimed at encouraging orphan drug research and development, frequently deters the pharmaceutical industry from showing any interest in this field. In 2019, the Indian government introduced the regulatory guidelines for gene therapy, 30 years after US conducted its first successful clinical trial. Despite not having incentives or other benefits, Indian pharmaceutical companies like Zydus Cadilla have received approval from foreign approval authorities and are selling such drugs and therapies in overseas markets (The Economic Times2021) India needs to come up with a policy allowing manufacturing of orphan drugs as soon as possible. With India's growing market and increasing urban population, the number of middle-class people who will be able afford the drugs is increasing. For those who cannot afford these drugs, government can devise health coverage schemes like the Mukhyamantri Amrutum Yojna in Gujarat and cover the costs of the drugs under it. It is high time that the Government addresses the issue, and frames a relevant regulation for encouraging the development of orphan drugs for rare diseases.

THE CURRENT SITUATION

The Policy states that the cost of treatment of rare diseases may vary from INR 10 lakhs to INR 1 crore on an annual basis. In addition to the treatment, some diseases require lifelong support. There are other associated overhead costs. The amount of 20 lakh sanctioned under the

Rashtriya Arogya Nidhi is barely enough to cover the costs of treating a rare disease in an average Indian household.

Currently, people are left at the mercy of a few NGOs and Foundations to help them collect the cost of treatment. As of August 2019, Takeda Pharmaceutical Company's programme has covered 199 patients from 13 countries including India, and Sanofi Genzyme has provided free drugs to more than 100 patients in India in the past 21 years (Salian 2021). In recent times, people are increasingly relying on crowdfunding platforms along with social media campaigns to gather funds for patients with rare diseases. This method has been successful in garnering funds for a lot of people. Recently, a whopping 16 crore was collected by a Hyderabad-based couple to procure a drug called Zolgensma for their three-year-old son who was suffering from Spinal Muscular Atrophy (Rao 2021). A similar story was of two-year-old boy Aayansh Gupta, whose parents raised 14.3 crores for a drug to treat his rare disease in November 2020 (Economic Times 2021).

With the success of crowdfunding-led initiatives, the Government took note of it and proposed crowdfunding as one of the solutions in the new Policy. The Central Government recently informed the Delhi High Court that a digital platform has been made operational for crowdfunding of treatment and medicines for rare diseases.

The private initiatives are leading the war, but without support from the Government, such solutions are not sustainable. Support from foundations, NGO's and crowdfunding-led initiatives, though extremely helpful, will not be accessible to all and is only a stop-gap solution. The treatment for rare diseases can take a financial toll on the suffering families. Recovery takes a long time after the treatment. This requires sustained medical and financial support to the patient, which can only be obtained with support from Government. There is a need to address the highlighted issues and come up with a more robust and inclusive policy in consultation with the State Governments. It needs to be ensured that this public health concern is addressed sustainably to provide respite to thousands of victims of rare diseases and their families in India.

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