Balancing patient need with public policy in rare diseases – A legal perspective

The landscape of rare diseases is a challenging cohort of medical conditions. The definition of a "rare" disease itself is debatable and could be country or region specific. Furthermore, it is a dynamic list that changes with advancements in research, new treatment modalities, and public awareness. Other than a few conditions, where there is considerable progress, the field is relatively nascent and largely unexplored.

From a legislative perspective, despite the challenges, the Indian Government, in a bold and positive step, sought to address the topic of rare diseases through the National Policy for Treatment of Rare Diseases (NPTRD) in July 2017 formulated by the Ministry of Health and Family Welfare, Government of India. However, its implementation was difficult because of the unaddressed gaps including the issue of cost-effectiveness of supporting such health interventions, the role of states, and regional differences in legislation and medical facilities. Some of these gaps were addressed after seeking public opinion from stakeholders, organizations, and states and were refined in the more recent National Policy for Rare Diseases 2021.[1]

There are still some unmet challenges that need to be highlighted. Firstly, defining a rare disease requires assessment of the prevalence of that condition in the community. This has seldom been performed in India owing to the cost of screening tests (including expensive genetic testing) and limited clinical trials. Secondly, treatment options are either unavailable, difficult to procure, or prohibitively expensive. A classic example would be the cost of voretigene neparvovec, sold under the brand name Luxturna, a gene therapy medication for the treatment of Leber congenital amaurosis (LCA) developed by Spark Therapeutics.[2] The cost of the one-time therapy is USD 850,000, which is over INR 5 crores, which puts it out of reach of most patients. Finally, the lack of epidemiological data in India on rare diseases makes it very difficult to assess the economic burden, morbidity, and mortality of these diseases, further widening the gap between demand and supply.[3]

It is interesting to note that the National Policy for Rare Diseases 2021[1] divides these diseases into three groups: Group 1: diseases amenable to a one-time curative treatment; Group 2: diseases requiring long-term or lifelong treatment, having a relatively lower cost of treatment, but with benefit; and Group 3: diseases for which definitive treatment is available, but with challenges like optimal patient selection for benefit, very high cost, or lifelong therapy. Unfortunately, no ophthalmological condition is explicitly mentioned in any of the groups, but there is a provision in the policy to update this list going forward.

As a socialist nation, we must endeavor to provide health care to all our citizens. Given our population and health-care access, this remains an aspirational goal rather than one that is pragmatically achievable. Besides, there are legal implications too. In a landmark case titled Mohd. Ahmed (Minor) vs. Union of India & Ors.,[4] the petitioner, a minor child suffering from Gaucher's disease and who was the only surviving child of his parents, having lost three siblings to the disease, approached the Delhi High Court seeking free and lifelong treatment. He submitted that health should be guaranteed under Article 21 of the Constitution as a Fundamental Right. The cost of the enzyme replacement therapy was about Rs. 600,000 per month, which was unaffordable for the family. The State argued that there were no "unlimited resources" at their disposal and provision of facilities cannot be "unlimited." The Court ruled that "health is not a luxury and should not be the sole possession of a privileged few" and termed it a violation of Article 21 and directed the Government to provide the child the required therapy at the All India Institute of Medical Sciences for as long as he required it. There are other cases that raise a similar concern in the context of rare diseases.

As health-care workers, scientists, physicians, and foremost, as citizens of India, it is time to introspect. What is the long-term solution for rare diseases in general and rare eye diseases in particular? Where do we strike the balance? The National Policy for Rare Diseases 2021 has a provision for a single-time payment of Rs. 20 lakhs for these conditions, but is this sustainable and more importantly scalable? Perhaps a holistic approach is required. Strengthening primary prevention, detecting high-risk couples before conception, and secondary prevention strategies like robust, low-cost, and universal prenatal screening and newborn screening are imperative. Early diagnosis on one hand and improving access to tertiary care and translational research on the other hand would be the need of the hour. The national policy is exploring alternate funding such as crowdsourcing, supporting low-cost indigenous research, and expanding the "orphan drug" definition for pharmaceuticals and industry to support. The road is long and hard, but the travel is inevitable. The diseases may be rare, but compassion and care combined with a more robust approach through science and technology must be commonplace.

Anand Vinekar, Chaitra Jayadev

Department of Ophthalmology, Narayana Nethralaya Eye Institute, Bengaluru, Karnataka, India.
E-mail: drchaitra@hotmail.com

References

1. Ministry of Health & Family Welfare. National Policy for Rare Diseases 2021. Government of India; 2021. Available from: https://main.mohfw.gov.in/sites/default/files/Final%20NPRD%2C%202021.pdf. [Last accessed on 2022 Apr 08].
2. Choudhury MC, Chaube P. Integrating rare disease management in public health programs in India: Exploring the potential of National Health Mission. Orphanet J Rare Dis 2022;17:43.
3. Nuzbrokh Y, Ragi SD, Tsang SH. Gene therapy for inherited retinal diseases. Ann Transl Med 2021;9:1278.
4. Available from: https://indiankanoon.org/doc/77985236/. [Last accessed on 2022 Apr 08].

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_905_22

Cite this article as: Vinekar A, Jayadev C. Balancing patient need with public policy in rare diseases – A legal perspective. Indian J Ophthalmol 2022;70:2584.