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Review Article

Equity for the rare: a review on India's rare disease challenges and policy responses

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ABSTRACT

Rare diseases, despite affecting a relatively small percentage of the population, impose significant health and economic burdens globally. India, with its vast genetic diversity and socio-cultural practices like consanguinity, faces unique challenges in managing rare diseases. The National Policy for Rare Diseases (NPRD) 2021 was introduced to streamline care, but major gaps persist in awareness, diagnosis, financing, and access. A scoping review was conducted using peer-reviewed literature, government reports, policy documents, and grey literature. Sources included Ministry of Health and Family Welfare, Indian Council of Medical Research, and advocacy organizations like Organization for Rare Diseases India. Data were analysed thematically across five domains: awareness, primary care, financial risk, health technology assessment, and policy responses. The review revealed significant deficits in rare diseases awareness among healthcare professionals and the public, with limited training and delayed diagnoses being common. Primary care lacks the resources for early genetic screening or effective referral systems. Financial burden remains high; treatments often exceed ₹1 crore, while financial aid caps at ₹50 lakhs. Health technology assessment methods are not adequately adapted for rare diseases, and orphan drug availability is hindered by regulatory and pricing barriers. While India has taken steps to improve rare disease care through the NPRD and policy reforms, critical barriers remain. Enhancing early diagnosis, revising health technology assessment frameworks for rare diseases, expanding local drug production, and integrating rare disease education into medical curricula are essential to achieving equitable and sustainable care for rare disease patients.

Keywords: Economic burden, Health burden, HTA, India, Rare diseases

INTRODUCTION

Rare diseases also known as "orphan diseases" are defined as conditions that affect only a small number of people predominantly based on the prevalence or incidence within a specific country or geographical region.[1] World Health Organization defines rare disease as those affecting fewer than 1 in 1,000 people.[2] However, definitions can differ across institutions and regions. For instance, the Organization for rare diseases India defines a rare disease as one affecting fewer than 100 individuals per 100,000 population, and an ultra-rare disease as affecting fewer than 2 individuals per 100,000 population.[3] This variation reflects the lack of a universally accepted definition of rare diseases. There are an estimated 6,000 to 8,000 rare diseases globally, affecting a significant number of people.[4-6] Notably, around 80% of rare diseases have a genetic basis, often manifesting early in life and requiring specialized care.[5] Worldwide, around 300 million people are affected with a rare disease, half of them being children, with around 30% likely to die by the age of five years.[7]

India, home to one of the most genetically diverse populations in the world with over 4,600 population groups, is uniquely positioned in the context of rare diseases.[8,9] Cultural practices of consanguineous marriages and endogamy within certain Indian sub-populations have

led to the accumulation of genetic traits and a higher prevalence of specific rare diseases.[10] Recognizing this burden, the Government of India introduced the National Policy for Rare Diseases (2021), which aims to improve access to diagnosis, treatment, and research for patients with rare conditions and has categorised the diseases as mentioned in Table 1.

Table 1: Categorisation of rare diseases according to national policy for rare diseases.

Group	Description	Examples
Group 1	Diseases that are amenable to one-time, high-cost curative treatment, often unaffordable for the poor. Includes organ transplants	Lysosomal storage disorders, adrenoleukodystrophy, severe combined immunodeficiency disorders, liver and renal transplantation
Group 2	Diseases requiring lifelong treatment using low-cost dietary/nutritional/hormonal supplements	Phenylketonuria, growth hormone deficiency, cystic fibrosis
Group 3	Diseases needing very high-cost lifelong treatment with significant challenges in patient selection	Gaucher's disease, Hunter's syndrome, Duchenne muscular dystrophy

Despite the substantial and growing burden of rare diseases, there remains a significant paucity of data on their hidden prevalence in India, particularly at the community and primary healthcare levels. Patients and families continue to face immense challenges even as advancements in genetic technologies offer potential solutions. Given the heterogeneity and fragmented nature of the available evidence, a scoping review approach is more appropriate than a systematic review. This review therefore adopts the Arksey and O'Malley framework (2005) for scoping reviews, enabling a comprehensive synthesis of challenges, policies, and opportunities related to rare diseases in India.[11] This review focuses on individuals and families affected by rare diseases in India, with particular emphasis on the barriers and challenges related to timely diagnosis, treatment, and long-term management within the broader setting of the Indian health system, including policy frameworks, healthcare infrastructure, financing mechanisms, and sociocultural factors.

The objectives of this review are to map the existing evidence on the key challenges hindering access to diagnosis and treatment for individuals living with rare disease in India, and to propose actionable strategies to address these barriers through strengthened policy frameworks, sustainable financing mechanisms, and integration within the broader health system.

METHODS

We conducted a scoping review to explore the experiences of individuals living with rare diseases in India, focusing on the key challenges they face in accessing timely diagnosis, treatment, and long-term care. Further, we also examined the range of national policies, strategic frameworks, and government-led initiatives that have been introduced to address these issues.

The review was guided by the methodology proposed by Arksey and O'Malley (2005), for mapping evidence on rare diseases within the context of the Indian healthcare system.

A literature search was conducted across multiple sources, including peer-reviewed academic databases (such as PubMed and Google Scholar), official government publications, policy documents from the Ministry of Health and Family Welfare, the Indian Council of Medical Research, and relevant non-governmental and international organizations working in the field of rare diseases.[2,12-16] Grey literature, including technical reports, press releases, and relevant updates from advocacy groups was also included. The search was not limited by publication time and only English language articles were reviewed with a focus on those most relevant to the Indian health system and policy context.

After discussing and resolving any divergence, two writers (AA and AS) independently retrieved data from the relevant papers. In order to resolve any differences in the study and data, authors SD and PH were involved. Following a full-text examination by AA and PH, the paper was excluded if the data was unclear. After the first search and publication separation, data was independently generated in an Excel file based on the title and abstract with summarization of findings conducted by AA and AS. Rayyan software was used to handle the articles. Thematic analysis was employed to categorize the findings into five domains: "Disease awareness", "Primary care", "financial risk", "HTA", "Policy incentives" and "special access programmes". A final review and validation of the synthesis were undertaken by PH.

RESULTS

Challenges in rare diseases

Disease awareness

Evidence highlights a significant lack of awareness and understanding of rare diseases among the public, healthcare professionals, and researchers in India. Nearly 43% of healthcare professionals had not encountered a single rare disease patient in their practice.[17] In line with the broader trends, around 29% of healthcare professionals and 31% of researchers were unaware of the time required to diagnose rare diseases, indicating limited exposure to

these conditions and gaps in understanding their clinical course.[18] Most people relied on research papers and social media to stay informed and over 75% of healthcare professionals had no formal training or institutional programs on rare diseases.[17,18] The lack of foundational knowledge was also evident with people being unfamiliar with the definition of rare diseases, their prevalence, or the population affected, often confusing rare diseases with more common conditions.[17] A clear gap between knowledge and practice is evident in the domain of genetic disorders—for instance, in a Kolkata slum, 53% of the eligible couples had knowledge about thalassemia, yet only 9.6% underwent premarital screening and just 0.67% accessed prenatal diagnosis.[19] Similarly, in Odisha, while awareness of sickle cell disease was relatively high, most individuals lacked understanding of inheritance risks and the importance of screening, particularly in tribal communities.[20] High-performance liquid chromatography, considered a standard for screening hemoglobinopathies due to its sensitivity and accuracy, is available in many health settings but remains underutilized, primarily due to stigma, fear of rejection in marriage, and lack of access to genetic counselling.[21] In a study in Western India, even among couples identified as high-risk, 99% proceeded with marriage despite counselling, opting instead for prenatal diagnosis in subsequent pregnancies.[22]

Primary care

Primary healthcare providers play a crucial role in improving the quality of life for patients and families affected by rare diseases. They facilitate timely referrals to specialists, coordinate multidisciplinary care, and support patients in accessing necessary services.[23] However, managing rare diseases at the primary care level presents distinct challenges, including delayed diagnoses and limited availability of information, expertise, and treatment options.[24]

Genetic testing and counselling are essential for early diagnosis and effective management of rare diseases, yet they are often inaccessible at the primary level due to the high cost of genetic screening technologies.[23] It is estimated that screening all newborns in India for just one congenital disorder would consume approximately 1.2% of the nation's total health budget, underscoring the financial burden of implementing such initiatives at a large scale.[25]

In terms of treatment, symptomatic management was most reported by healthcare professional, reflecting lack of access to advanced therapies such as genetic or biologic treatments. Over 80% of healthcare professionals surveyed indicated that they lacked the necessary resources for diagnosis and treatment.[17]

Financial risk

Rare diseases pose a significant financial burden globally, with high-income countries often spending hundreds of thousands to millions of dollars per patient annually, supported largely by insurance or state subsidies. In upper middle income countries like China, treatment costs still account for a large portion of household income, with out-of-pocket expenses reaching over 40%. In contrast, India faces a severe gap—while treatments for conditions like spinal muscular atrophy can cost ₹2–5 crores per patient, government assistance under the National Policy for Rare Diseases is limited to ₹50 lakhs, leaving most of the financial burden on families. This disparity highlights the urgent need for more sustainable funding and inclusion mechanisms in India's rare disease policy.[26]

Out-of-pocket expenditure for the treatment of rare diseases can be extraordinarily high, often making access to care unaffordable for many families. For example, in the case of idiopathic pulmonary fibrosis, a rare and progressive lung disease, the only available treatment until recently was a lung transplant, which is both costly and invasive. This condition carries a 50% survival rate at just three years, underscoring the urgency and severity of the disease. The introduction of newer therapies such as Pirfenidone has offered less invasive options, but such treatments still remain expensive and largely inaccessible without financial support mechanisms, highlighting the critical need for comprehensive funding policies for rare diseases.[26]

The financial assistance scheme for poor patients suffering from rare diseases under Rashtriya Arogya Nidhi aims to support individuals living below the poverty line who are affected by specified rare diseases, enabling them to access treatment at government hospitals and institutes equipped with super-specialty facilities. In this regard, the Ministry of Health & Family Welfare launched the National Policy for Rare Diseases in March 2021. Based on the recommendations of the Central Technical Committee for Rare Diseases, a total of 63 rare diseases have been included under this policy. Eligible patients can receive financial assistance of up to ₹50 lakhs per case for treatment at the designated Centres of Excellence for Rare Diseases, as notified under the policy.[14,15]

Nevertheless, extremely high cost of treatment for rare disease remains the biggest challenges patients in India. For example, treating Lysosomal Storage Disorders with Enzyme Replacement Therapies can cost anywhere between 20 lakhs to 2.5 crores per year. In some cases, like spinal muscular atrophy, a single dose of gene therapy such as Zolgensma can cost as much as Rs. 18 crores.[24] Although the National Policy for Rare Diseases provides financial aid at notified Centres of Excellence for Rare Diseases, this amount is often not enough, forcing families to take loans or turn to crowdfunding. India already produces the active pharmaceutical ingredients for more than 400 FDA-approved orphan drugs, but the majority of these medications are not readily available and are not reasonably priced due to which patients still have to pay high prices.[27] This is because the APIs are exported and then re-imported as finished medicines, which adds extra costs like 10% import duties, packaging, and shipping. In

addition, many of these drugs need to be kept at a specific temperature during transport, which requires expensive cold-chain packaging. All these factors increase the overall financial burden on families seeking treatment.[28]

Health technology assessment

Health technology assessment encounters several critical challenges in conducting economic evaluations for rare diseases. One of the primary limitations is the lack of reliable epidemiological data, including accurate estimates of incidence, prevalence, and long-term health outcomes, which impedes precise assessment of disease burden.[29] Moreover, lack of clear prioritization of decision problems by the government and the absence of standardized health technology assessment methodologies tailored to rare diseases creates inconsistencies in evaluation approaches, further complicating economic assessments. The exceptionally high cost of treatment, alongside the lack of effective comparators, makes it particularly difficult to satisfy conventional cost-effectiveness thresholds, such as achieving incremental cost-effectiveness ratios within acceptable willingness-to-pay limits.[30] These factors collectively challenge the application of conventional health technology assessment methodologies in this context.

Policy incentives and special access programmes

Conducting research and clinical trials for rare diseases in India faces several challenges. Patients are rarely included in clinical drug trials due to a combination of factors, including a shortage of trained rare disease specialists, which often leads to delays in diagnosis or even misdiagnosis. Additionally, regulatory hurdles make it difficult to approve and conduct trials for experimental drugs within the country.

The high cost of importing these investigational therapies further limits their availability for clinical use. These barriers collectively restrict patient participation in clinical research and delay access to potentially life-saving treatments.[18,24]

To address these persistent challenges, the Government of India introduced a series of policy reforms aimed at strengthening the rare disease ecosystem. Figure 1 shows the timeline of the National Policy for Rare Diseases in India (2017-2021).

The Ministry of Health and Family Welfare, Government of India, under the National Health Mission launched the Rashtriya Bal Swasthya Karyakram to promote early diagnosis and interventions for 4 Ds - defects at birth, diseases, deficiencies and development delays, spanning 32 common health conditions for early detection and free treatment and management, including surgeries at tertiary level.[12] Additionally, The ICMR National Registry for Rare and Other Inherited Disorders, launched in November 2019 in, aims to systematically collect comprehensive data on the demographics, clinical presentation, natural history, and treatment outcomes of specific rare diseases.[13]

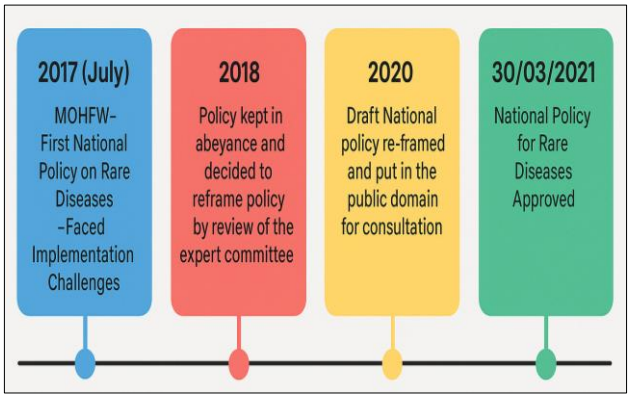

Figure 1: Timeline of the national policy for rare diseases in India (2017-2021).

Under the National Policy for Rare Diseases, financial assistance for conditions classified under group 1 is provided through the Rashtriya Arogya Nidhi, a central government fund. This support offers up to ₹50 lakh as a one-time grant for eligible treatments—an increase from the earlier cap of ₹20 lakh. The beneficiaries of this scheme include individuals from economically disadvantaged backgrounds who face severe financial hardship due to high healthcare costs. Importantly, eligibility is not limited to those officially below the poverty line (BPL); instead, it extends to the bottom 40% of the population as identified in the Ayushman Bharat PM-JAY database. Although the funding is not sourced from the PM-JAY scheme itself, the beneficiary list is drawn from its eligibility criteria. The government has initiated the launch of 'Made in India' drugs to address treatment gaps and significantly reduce the cost of therapies for rare diseases, particularly for conditions such as tyrosinemia type 1, Gaucher's disease, Wilson's disease, and Dravet/Lennox-Gastaut syndrome. This effort is part of the broader "Make in India" initiative and aims to enhance the affordability and accessibility of treatments within the rare disease space[32]. Supporting this approach is the landmark Supreme Court judgment in the Novartis versus Union of India, where patent protection for the cancer drug Glivec was denied. The ruling reaffirmed India's commitment to prioritizing public access to essential medicines over prolonged patent monopolies, especially in cases involving minor modifications to existing drugs.[33] To strengthen the infrastructure for diagnosis, counselling, and treatment of rare diseases, the Government of India has established two types of designated institutions under the National Policy for Rare Diseases.

Nidan Kendra's

These centres, established by the Department of Biotechnology under the Unique Methods of Management and treatment of inherited disorders (UMMID) initiative,

play a vital role in rare disease management. Nidan Kendra's provide genetic testing,

screening, and counselling services, while treatment, where available, is delivered under the supervision of a recognized Centre of Excellence (CoE). The CoEs undertake a wide range of activities including patient treatment, antenatal and neonatal screening, advanced diagnostics such as cytogenetic, molecular and metabolic testing, and prevention through prenatal screening and awareness.

They also focus on capacity building through education and training at all levels and lead research efforts aimed at developing affordable diagnostic and therapeutic solutions.[34] The list of Nidan Kendra's established across India is provided in Table 2.

Centres of excellence

Twelve institutions across India have been designated as Centres of Excellence to provide comprehensive care for rare diseases, as detailed in Table 3. These centres of Excellence are responsible for developing standard operating protocols, enabling early diagnosis, streamlining care pathways, and improving quality of life for affected individuals.

Table 2: Nidan Kendra's established in India

S. no.	Nidan Kendra	Location
1	Lady Hardinge Medical College (LHMC)	Delhi
2	Nizam's Institute of Medical Sciences (NIMS)	Hyderabad, Telangana
3	All India Institute of Medical Sciences (AIIMS)	Jodhpur, Rajasthan
4	Army Hospital Research and Referral	Delhi
5	Nil Ratan Sircar Medical College and Hospital	Kolkata, West Bengal

Recommendations for strengthening rare disease management

To enhance the management and treatment of rare diseases, a comprehensive and integrated strategy is essential. Raising awareness among primary care physicians and the general public plays a pivotal role in facilitating early diagnosis and timely intervention, alongside the implementation of robust screening programs for high-risk populations. Strengthening diagnostic and therapeutic capabilities through improved access to advanced technologies and sustained investment in research and development is also vital.

From an economic standpoint, it is important to revise health technology assessment frameworks to better accommodate the specific challenges posed by rare diseases, with a focus on long-term quality of life outcomes. Additionally, promoting domestic production of orphan drugs can contribute to greater sustainability and reduce reliance on external sources in rare disease care. A summary of challenges and possible solutions at immediate, intermediate and long-term levels is provided in Table 4.

To strengthen rare disease management, a structured three-tiered model, as shown in the figure 2, enables timely diagnosis, treatment, and follow-up. At the primary level, family history screening and preconception counselling help identify at-risk individuals. The secondary level focuses on newborn screening, confirmatory diagnostics, and initial management. For complex cases, the tertiary level provides specialist care, definitive treatment, registry entry, and telehealth follow-up.

Education, training, and community awareness are key enablers. Integrating this approach with supportive policies like indigenous orphan drug production, flexible health technology assessment, and research and development investment can improve outcomes and reduce delays in rare disease care.

Table 3: Centres of excellence established in India.

S. no.	Centre of excellence	Location
1	All India Institute of Medical Sciences (AIIMS)	New Delhi
2	Maulana Azad Medical College	New Delhi
3	Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGI)	Lucknow, Uttar Pradesh
4	Postgraduate Institute of Medical Education and Research (PGIMER)	Chandigarh
5	Centre for DNA Fingerprinting & Diagnostics (with NIMS)	Hyderabad, Telangana
6	Seth GS Medical College and KEM Hospital	Mumbai, Maharashtra
7	Institute of Postgraduate Medical Education and Research (IPGMER)	Kolkata, West Bengal
8	Centre for Human Genetics (in collaboration with IGH)	Bengaluru, Karnataka

Continued.

Table 4: Possible solutions for various challenges associated with rare diseases.

Challenges	Immediate solutions	Intermediate solutions	Long-term solutions
Primary care	Targeted awareness for primary care physicians and general public, adequate newborn screening for vulnerable groups	Availability of genetic disorder diagnostic facilities	Investment in research and development
Economic evaluations	Relaxations for HTA of rare diseases, less stringent clinical and economic evidence, long-term QoL over short-term	Development of a structured framework for HTA of rare diseases	Strengthen National Disease registry, fixing a budget impact threshold for resource allocation
Healthcare financing and pricing	Subsidized access, rare disease funds	Conditional reimbursement	Promoting domestic manufacturers for RD drugs

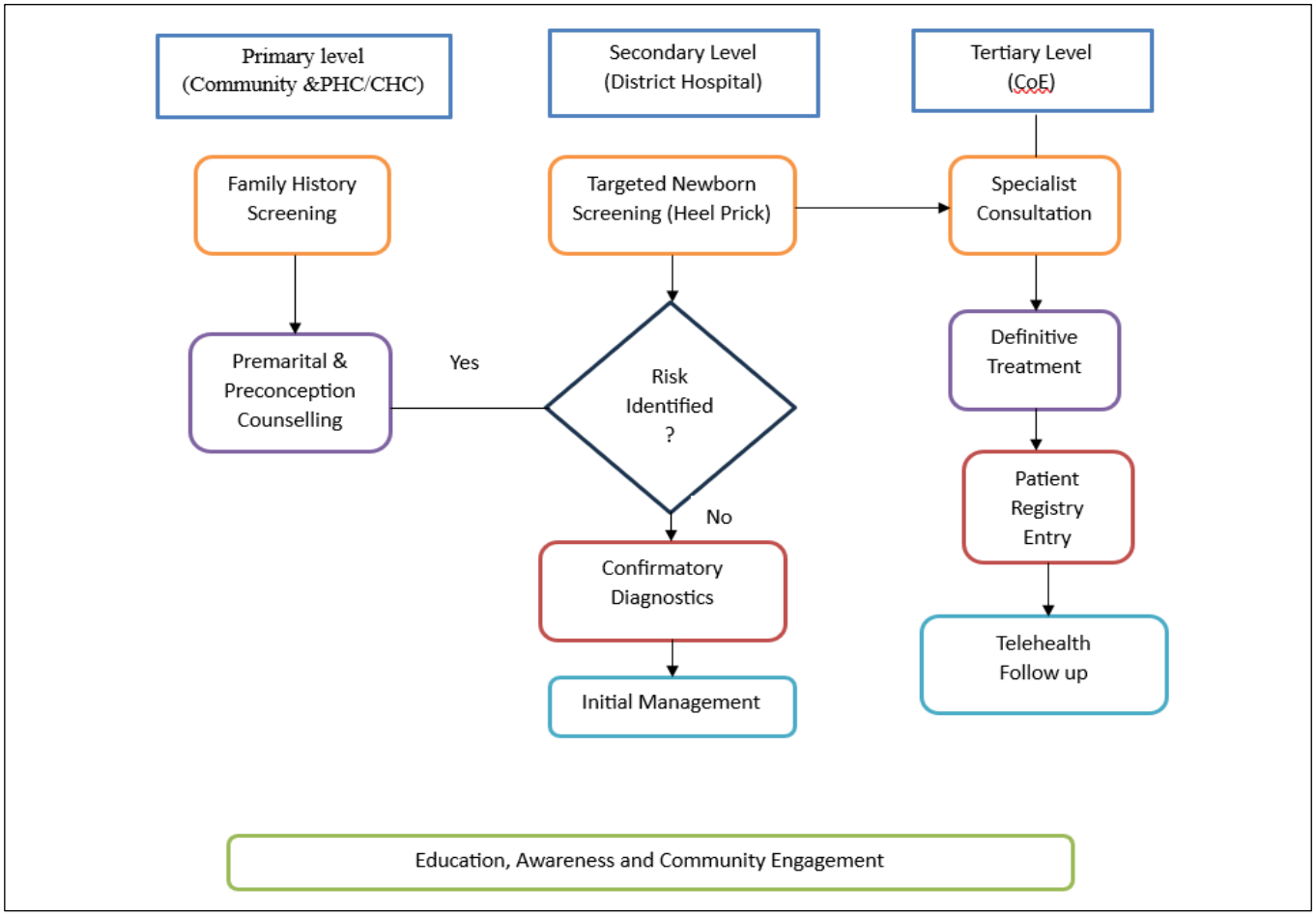

Figure 2: Three-tiered model for prevention, treatment and management of rare diseases.

DISCUSSION

The findings of the review highlight a critical and persistent gap in the awareness and understanding of rare disease within the Indian context, particularly among medical students and frontline health care providers. Despite advances in the public health infrastructure, rare disease continues to receive limited attention. The lack of formal training, insufficient diagnostic support and minimal public discourse contributes to delayed diagnosis and sub-optimal care for individuals living with rare disease. While these challenges are especially evident in India, they are reflective of the global trends. Countries reported evident gap in foundational knowledge on rare diseases and highlighted urgent need to strengthen rare disease related education by integrating additional courses in medical curriculum and by providing continuing education courses focused on rare diseases.[35,36] In addition to educational barriers, the capacity of primary care systems to manage rare diseases remains underexplored. A study from the U.S. found that only 1.6% visits to primary care providers involved patients with rare disease.[37] This low representation raises questions on the level of preparedness of primary care providers in managing rare disease. However, several high-income countries have implemented models that aim to strengthen the role of primary care in rare disease management. For instance, the United Kingdom integrates rare disease awareness into

general practice through continuing medical education and referral networks such as the European Reference Networks, which provide guidance to frontline providers on managing complex and rare cases.[6] Similarly, countries like Canada and Australia have embedded genetic counsellors and rare disease coordinators within community health settings to assist primary care physicians in early detection and patient navigation.[38,39] These systems ensure that primary care physicians are not only gatekeepers but also informed actors in the care continuum. In contrast, India's primary care system lacks such integrated support and training, limiting its ability to serve as a first point of contact for rare disease patients. Empowering Indian primary care providers through structured training, teleconsultation access to specialists, and referral protocols could significantly reduce diagnostic delays and promote equity in health outcomes.

On the policy front, countries are increasingly adapting their Health Technology Assessment frameworks to respond to the unique challenges posed by rare diseases and orphan drugs. Conventional Health Technology Assessment methodologies centered on cost-effectiveness thresholds, quality-adjusted life years, and population-level impact are unable to adequately capture the value of interventions for small, dispersed patient populations with high unmet needs. In response, several countries have implemented specialized health technology assessment processes that incorporate broader social, ethical, and equity considerations into decision-making. The National Institute for Health and Care Excellence in the United Kingdom has developed the specialized health technology assessment process to evaluate orphan drugs and ultra-orphan drugs and a highly specialised technologies programme, designed specifically for evaluating treatments for very rare conditions.[40] In Thailand, the Health Intervention and Technology Assessment Program has made significant strides in assessing interventions for rare diseases, particularly in the context of the country's Universal Health Coverage framework. Recognizing that conventional incremental cost-effectiveness ratio thresholds may exclude high-cost but essential treatments for rare conditions, health intervention and technology assessment program emphasizes contextualized decision-making, including budget impact, ethical implications, and stakeholder input. Thailand has successfully included rare disease treatments such as enzyme replacement therapy for Gaucher disease and congenital adrenal hyperplasia into its benefit package after stakeholder and public consultations.[41]

As India advances its commitment to addressing rare diseases through the National Policy for Rare Diseases (2021), it must prioritize the creation of an inclusive, patient-centered healthcare ecosystem. Strengthening early diagnosis, expanding access to affordable therapies, enhancing primary care capacity, promoting widespread awareness and sustainable financing mechanisms to reduce the heavy out-of-pocket burden currently borne by patients and families are essential steps towards achieving this goal.

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