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Strengthening India's Rare Disease Ecosystem

A policy roadmap for early diagnosis, equitable access, and Better outcomes

EARLY DIAGNOSIS

EQUITABLE ACCESS

BETTER OUTCOMES

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Executive Summary

Rare diseases are uncommon individually, but collectively affect an estimated 300 million people worldwide across more than 7,000 identified conditions.[1] Roughly 80% of them are genetic in origin, about 70% present in childhood, and 95% still lack an approved treatment. The average time to an accurate diagnosis is estimated at 4.8 years globally[2], a delay that is often far longer in low- and middle-income countries, including India.

300M+ **People living with a rare disease worldwide**	**7,000+** **Distinct rare diseases identified**	**4.8 years** **Average time to an accurate diagnosis**	**95%** **of rare diseases have no approved treatment**

India carries a substantial share of this global burden.[3] Estimates suggest tens of millions of Indians live with a rare disease, yet the country still lacks a nationally validated epidemiological baseline, a fully operational patient registry, and a self-sufficient domestic ecosystem for diagnostics and orphan drug manufacturing. The National Policy for Rare Diseases (NPRD), 2021 marked an important turning point[4] introducing a three-tier disease classification, financial assistance of up to ₹50 lakh per patient, a crowdfunding mechanism, and a growing network of designated Centres of Excellence (CoEs), which has expanded from an initial 8 institutions to 15 as of 2026.[5] Implementation, however, has been uneven: patient advocacy groups continue to report delays in fund disbursement, limited geographic reach of CoEs, and slow progress on the national registry.

This paper examines India's current rare disease landscape, reviews the policy and regulatory frameworks of six jurisdictions with mature rare disease ecosystems that is the United States, the European Union, the United Kingdom, Japan, Australia, and Singapore and examines the lessons most transferable to India's context.

It identifies eight structural policy gaps and proposes a six-pillar roadmap: (1) earlier diagnosis, (2) improved and sustainable access, (3) strengthened research and real-world

[1]The Lancet Global Health, 'The Landscape for Rare Diseases in 2024' (Editorial), Vol. 12, 2024.
[2]The Lancet Global Health, 'The Landscape for Rare Diseases in 2024' (Editorial), Vol. 12, 2024.
[3]Rare Diseases International / EURORDIS, 'New Scientific Paper Confirms 300 Million People Living With a Rare Disease Worldwide', 2019.
[4]Ministry of Health & Family Welfare, Government of India, National Policy for Rare Diseases, 2021.
[5]Indian Organization for Rare Diseases (IORD), 'India Expands Rare Disease Centres of Excellence Network to 15 Under NPRD 2021', rarediseases.in.

evidence, (4) domestic innovation incentives, (5) patient-centric and decentralised care (6) stronger governance and coordination.

The following ten priority recommendations are proposed to strengthen India's rare disease policy framework and its implementation.

Ten Priority Recommendations

1. Expand the National Rare Disease Registry into a mandatory, interoperable data platform linked to all Centres of Excellence.
2. Pilot phased newborn screening for priority of treatable Group 1 and Group 2 conditions, beginning in states with existing screening infrastructure.
3. Protect dedicated funding under the NPRD and simplify the approval and fund-release process to ensure timely access to the ₹50 lakh financial assistance for eligible patients.
4. Introduce managed-access and outcome-based reimbursement agreements for high-cost rare disease therapies, linking public funding to demonstrated patient outcomes and sharing financial risk between government and manufacturers, drawing on models adopted in the UK and Australia.
5. Extend the Ayushman Bharat / PM-JAY insurance architecture to include defined rare disease treatment packages.
6. Create fiscal incentives that is tax credits, accelerated approvals, and reduced regulatory fees for domestic orphan drug development and manufacturing, building on the Production Linked Incentive Scheme.
7. Double the number of Centres of Excellence over five years with at least one in every state and formalise referral pathways from district hospitals.
8. Strengthen genetic counselling across the care continuum by integrating structured rare-disease and genetic counselling training into medical education and continuing professional development, while expanding premarital and pre-conception counselling and targeted carrier screening for individuals and couples at increased genetic risk.
9. Establish a national inter-ministerial coordinating body for rare diseases, with annual public reporting on NPRD outcomes.
10. Establish structured collaborations among government, industry, research institutions, and patient organisations to collect long-term clinical data (natural history studies) and maintain repositories of patient biological samples (biobanks) to support rare disease research, diagnosis, and therapy development.

Collectively, these measures would help India transition from a foundational policy framework to an integrated rare disease ecosystem — reducing diagnostic delays, improving

equitable access to care, and strengthening domestic research, innovation, and manufacturing for long-term sustainability.

Introduction

What are rare diseases?

A rare disease is generally defined as a condition affecting a small proportion of the population, though the exact threshold varies by jurisdiction: the European Union uses a prevalence of no more than 5 in 10,000 people, the United States defines rarity as fewer than 200,000 affected individuals nationally, and several Asian regulators use a threshold of roughly 1 in 10,000. India does not yet have an official epidemiological definition of a rare disease, a gap acknowledged by the National Policy for Rare Diseases (NPRD), 2021.

Collectively, more than 7,000 distinct rare diseases have been identified, and approximately 300 million people live with one worldwide[6]. Around 80% of rare diseases have a genetic origin, and about 70% of these present during childhood. Roughly 95% of rare diseases have no approved treatment, and the average time to reach an accurate diagnosis is about 4.8 years[7], a period during which patients frequently see multiple specialists, undergo repeated misdiagnosis and a large share of paediatric cases do not survive to receive a diagnosis at all.

Why are rare diseases a growing policy priority?

Rare diseases have moved from a niche clinical concern to a mainstream health-system and industrial-policy issue for three converging reasons.

1. Advances in genomic sequencing, gene therapy and cell therapy are making an increasing number of rare conditions treatable, and in some cases potentially curable making timely and equitable access to these therapies an increasingly important policy priority.
2. As countries advance towards universal health coverage, health systems in emerging economies are increasingly expected to accommodate high-cost, low-volume therapies within their financing and service delivery frameworks.[8]
3. Pharmaceutical and biotechnology innovation is increasingly concentrated in specialty and orphan indications, making rare disease policy a determinant of where global R&D investment, clinical trials, and manufacturing capacity are located.

[6]The Lancet Global Health, 'The Landscape for Rare Diseases in 2024' (Editorial), Vol. 12, 2024.
[7]The Lancet Global Health, 'The Landscape for Rare Diseases in 2024' (Editorial), Vol. 12, 2024.
[8]World Health Organization Executive Board, 'Rare Diseases: A Global Health Priority for Equity and Universal Health Coverage', EB156 documentation, apps.who.int.

In India, where a large population translates into a significant absolute burden of rare diseases, and where the genomics and biotechnology sectors are expanding alongside ambitions to become a global hub for affordable innovation, rare disease policy has implications that extend beyond patient care to the broader health and innovation ecosystem.

Rare Disease in India: The Current Landscape

Epidemiology and Disease Burden

India lacks a validated national prevalence estimate for rare diseases; the NPRD document itself notes the absence of the epidemiological data needed to define rare diseases in terms of prevalence[9]. Commonly cited estimates suggest tens of millions of Indians live with a rare disease, and India's National Policy for Rare Diseases currently formally lists 63 rare diseases on the recommendation of its Central Technical Committee for Rare Diseases (CTCRD)[10]. This absence of a robust denominator is itself a policy gap: without reliable prevalence data, resource planning, budget allocation, and drug-pricing negotiations all proceed on an uncertain footing.

Illustrative Disease Areas

The rare diseases most visible in India's policy and clinical discourse span several categories: lysosomal storage disorders (Gaucher disease, Pompe disease, Fabry disease, Mucopolysaccharidosis types I and II), neuromuscular disorders (Spinal Muscular Atrophy, Duchenne Muscular Dystrophy), inherited metabolic and haematological disorders (Wilson's disease, Hypophosphatasia, Haemophilia, Thalassaemia), and immune or congenital conditions such as Osteopetrosis and Cystic Fibrosis. Sickle Cell Disease, while classified separately in India's public health strategy because of its comparatively higher prevalence in tribal and specific regional populations, shares many of the same diagnostic and access challenges as the rarer conditions above.

The Diagnostic Landscape

India's diagnostic gap has several interlocking causes: limited newborn and antenatal screening coverage outside a small number of states and private programmes; a shortage of accredited genetic-testing and molecular-diagnostics laboratories, most of which are concentrated in a handful of metropolitan centres; low awareness of rare disease presentations among primary-care and even many specialist physicians, who may not

[9]Ministry of Health & Family Welfare, Government of India, National Policy for Rare Diseases, 2021.
[10]Organization for Rare Diseases India (ORDI), 'List of Rare Diseases as per National Policy of Rare Diseases, 2021', ordindia.in.

consider a rare genetic cause until common differentials are exhausted; and pronounced urban–rural disparity in access to paediatric geneticists, metabolic specialists, and genetic counsellors. Together, these factors contribute to prolonged diagnostic delays in India, with many families experiencing years of unexplained symptoms, multiple hospital admissions, and repeated testing before receiving a definitive diagnosis.

The Treatment Landscape

For the minority of rare diseases with an approved therapy, access in India is constrained by near-total dependence on imported enzyme-replacement therapies, biologics, and increasingly gene and cell therapies, most of which are developed and manufactured abroad and priced in hard currency. Out-of-pocket costs for lifelong therapies such as enzyme replacement can run into tens of lakhs of rupees annually placing them beyond the reach of all but a small share of affected families even where treatment exists. Domestic manufacturing of orphan drugs remains limited, though government schemes such as the Production Linked Incentive (PLI) Scheme for Pharmaceuticals have begun to extend fiscal incentives to this category.[11]

Policy and Regulatory Landscape in India

The National Policy for Rare Diseases (NPRD), 2021

India's rare disease policy journey began with a draft framework prepared in 2017 which was placed on hold pending resolution of questions on disease criteria, cost-sharing, and eligible beneficiaries. An expert committee constituted in 2018 submitted its recommendations in January 2021 and the Ministry of Health and Family Welfare formally notified the National Policy for Rare Diseases on 31 March 2021[12]. The NPRD introduced several structural features that remain the backbone of India's rare disease framework today:

Disease Categorisation

- **Group 1 —** Disorders amenable to one-time curative treatment, such as certain conditions treatable through haematopoietic stem cell transplantation or organ transplantation.
- **Group 2 —** Diseases requiring long-term or lifelong treatment, but at a relatively lower cost, where a clear clinical benefit has been documented in the literature and requires periodic surveillance.

[11]Press Information Bureau, Government of India, 'Initiatives by the Government for Treatment of Rare Diseases', pib.gov.in.
[12]Organization for Rare Diseases India (ORDI), 'List of Rare Diseases as per National Policy of Rare Diseases, 2021', ordindia.in.

- **Group 3 —** Diseases for which definitive treatment exists but which involve very high, lifelong treatment costs and complex patient-selection considerations — the category into which most enzyme-replacement and gene therapies fall.

Financial Assistance

The policy initially provided financial support of up to ₹20 lakh per patient under the Rashtriya Arogya Nidhi (RAN) umbrella scheme for Group 1 conditions, restricted to treatment at government tertiary hospitals and to beneficiaries meeting the Pradhan Mantri Jan Arogya Yojana (PM-JAY) eligibility criteria then extending coverage to roughly 40% of the population rather than only those below the poverty line. In May 2022, this assistance ceiling was raised to ₹50 lakh per patient across all three disease groups, payable for treatment at any designated Centre of Excellence, outside the RAN umbrella scheme.

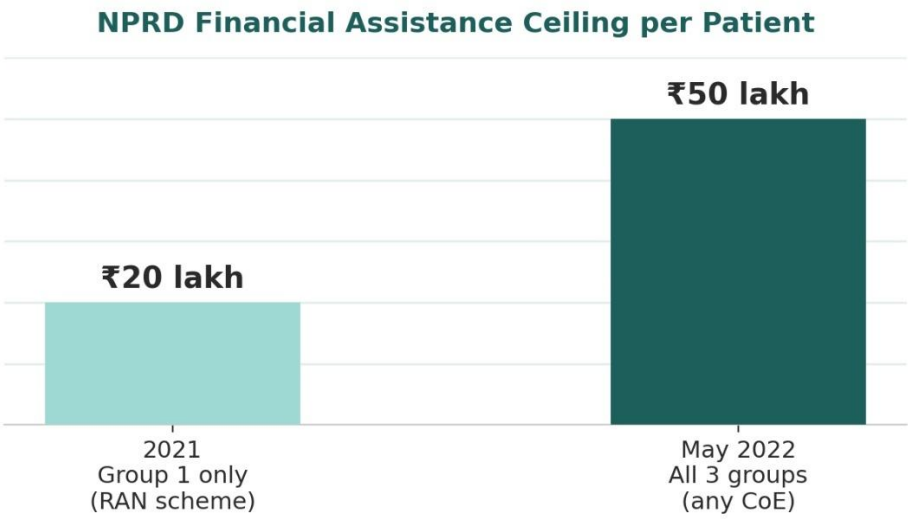

Figure 1: Growth in NPRD Financial Assistance Ceiling

Source: Ministry of Health & Family Welfare, National Policy for Rare Diseases, 2021

Crowdfunding, Registry, and Research Provisions

The NPRD also introduced a voluntary crowdfunding mechanism a digital platform intended to allow individual and corporate donors to contribute toward patients' treatment costs alongside provisions to promote indigenous research, local manufacturing of rare disease drugs at affordable prices, and the creation of a national, hospital-based Rare Disease Registry coordinated by the Indian Council of Medical Research (ICMR), intended to close India's epidemiological data gap over time.[13]

[13]Organization for Rare Diseases India (ORDI), 'List of Rare Diseases as per National Policy of Rare Diseases, 2021', ordindia.in.

Centres of Excellence

The NPRD designates a network of tertiary government hospitals as Centres of Excellence (CoEs), responsible for antenatal, neonatal, and high-risk screening, diagnosis and treatment; and research into low-cost diagnostics and therapeutics for rare diseases[14]. The network began with 8 institutions and has since been expanded in stages to 12 by mid-2025 and to 15 by 2026, with the most recent additions being the Regional Institute of Medical Sciences (RIMS), Imphal; AIIMS Patna; and Assam Medical College, Dibrugarh, alongside long-standing CoEs such as AIIMS New Delhi, PGIMER Chandigarh, SGPGIMS Lucknow, and CDFD/NIMS Hyderabad[15]. Even at 15 institutions, however, coverage remains concentrated, leaving many states without a designated centre and requiring long-distance travel for diagnosis and follow-up care.

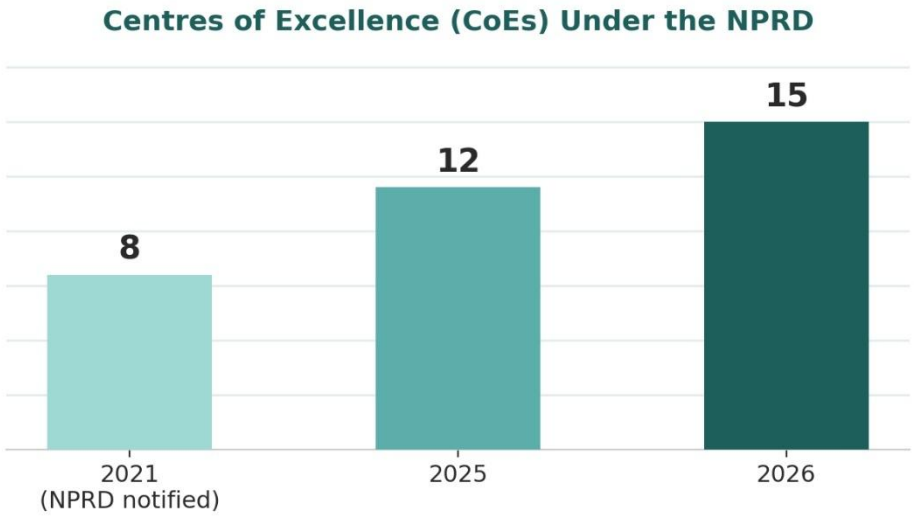

Figure 2: Expansion of the CoE Network (2021-2026)

Source: Indian Organization for Rare Diseases (IORD), 2026

Regulatory Framework

India's drug regulatory framework, administered by the Central Drugs Standard Control Organisation (CDSCO) under the New Drugs and Clinical Trials Rules, 2019, provides several regulatory flexibilities that can facilitate access to orphan drugs. These include waivers of local clinical trial requirements for drugs already approved in specified foreign jurisdictions, accelerated approval pathways for therapies addressing unmet medical needs, and compassionate-use provisions for eligible patients. While these mechanisms can support the

[14]Organization for Rare Diseases India (ORDI), 'List of Rare Diseases as per National Policy of Rare Diseases, 2021', ordindia.in.

[15]Indian Organization for Rare Diseases (IORD), 'India Expands Rare Disease Centres of Excellence Network to 15 Under NPRD 2021', rarediseases.in.

timely approval and availability of orphan drugs, India does not yet have a dedicated orphan drug regulatory framework comparable to those in jurisdictions such as the United States, the European Union, or Japan, where orphan drugs are governed through tailored designation, approval, and incentive pathways.

Funding Mechanisms Beyond NPRD

Beyond the NPRD's own financial assistance, rare disease patients in India may draw on a patchwork of additional mechanisms: state-specific health schemes (several states, including Kerala, Tamil Nadu, and Maharashtra, have supplemented central assistance with their own programmes); corporate social responsibility (CSR) contributions channelled through hospitals and patient organisations; and, increasingly, private and public crowdfunding platforms. The absence of a rare-disease-specific insurance product, and the exclusion of most high-cost orphan therapies from standard Ayushman Bharat/PM-JAY packages, means that financing remains fragmented and unpredictable for most families.

Key Challenges

1. Diagnosis

- Delayed diagnosis due to limited physician awareness and referral gaps.
- Very limited public newborn-screening coverage beyond a small panel of conditions in a few states.
- Shortage of accredited genetic-testing and next-generation-sequencing laboratories outside major metros.

2. Access

- High absolute cost of imported orphan therapies relative to household incomes.
- Geographic inequity: 15 CoEs cannot adequately serve a population spread across 28 states and 8 union territories.
- Reported delays in NPRD fund disbursement, which patient groups have flagged repeatedly since 2021.

3. Financing and Insurance

- No dedicated, ring-fenced rare disease insurance product; most Group 3 therapies fall outside PM-JAY package limits.
- Reliance on a discretionary ₹50 lakh assistance ceiling that may not cover a lifetime of Group 3 therapy costs.
- Limited use, so far, of internationally proven risk-sharing or outcome-based payment models.

4. Research and Evidence

- Few India-specific natural-history studies, which limits the local evidence base for regulatory and reimbursement decisions.
- A national registry that remains under development, constraining epidemiological planning and clinical trial recruitment.
- Limited India-based clinical trial activity for rare disease therapies relative to disease burden.

5. Domestic Manufacturing and Innovation

- Heavy dependence on imported biologics and gene therapies, with associated foreign-exchange and supply-chain exposure.
- A nascent, rather than mature, domestic orphan-drug development ecosystem, despite PLI-scheme incentives.

6. Data and Coordination

- No single, interoperable data system linking CoEs, insurers, and the Ministry of Health and Family Welfare.
- Fragmented governance across the Department of Health Research, ICMR, CDSCO, and state health departments.

Global Best Practices

Several jurisdictions have established comprehensive rare disease ecosystems providing useful lessons for India.

1. United States — Incentivising Innovation Through Market Exclusivity

The U.S. Orphan Drug Act of 1983 remains the template most other jurisdictions have adapted. It created two core mechanisms: FDA orphan-drug designation for products addressing conditions affecting fewer than 200,000 Americans, and a resulting seven-year period of market exclusivity for the approved orphan indication.[16] Designated products also receive a tax credit (historically up to 25–50% of qualifying U.S. clinical trial costs), a waiver of the substantial Prescription Drug User Fee Act (PDUFA) application fee, and exemption from mandatory paediatric study requirements under the Paediatric Research Equity Act[17]. Since 1983, the FDA has approved several hundred orphan-designated products estimates in the literature range from roughly 380 to over 500 depending on the counting methodology and

[16]U.S. Congressional Research Service, 'The Orphan Drug Act: Legal Overview and Policy Considerations', congress.gov, IF12605.
[17]U.S. Congressional Research Service, 'The Orphan Drug Act: Legal Overview and Policy Considerations', congress.gov, IF12605.

cut-off year a substantial increase from the handful approved in the years before the Act[18]. Evaluations of the policy note a genuine trade-off: while exclusivity has clearly expanded the pipeline of orphan therapies, it has also been associated with sustained high pricing, since exclusivity in some cases outlasts the underlying patent protection.[19][20]

2. European Union — Centralised Designation With Reinvestment Incentives

EU Regulation (EC) No. 141/2000 established a Community-wide procedure for orphan designation available to products treating conditions affecting no more than 5 in 10,000 people in the EU or those that would not otherwise generate a sufficient return to justify investment.[21] Designated products can receive ten years of market exclusivity (reducible to six years if the product no longer meets the criteria for orphan status), fee reductions, protocol assistance from the European Medicines Agency, and access to the centralised marketing-authorisation procedure across all member states simultaneously.[22][23] The EU further operates European Reference Networks (ERNs) virtual networks connecting specialist centres across member states for specific rare disease groups allowing cross-border pooling of clinical expertise for conditions too rare to sustain expert capacity in any single country.[24]

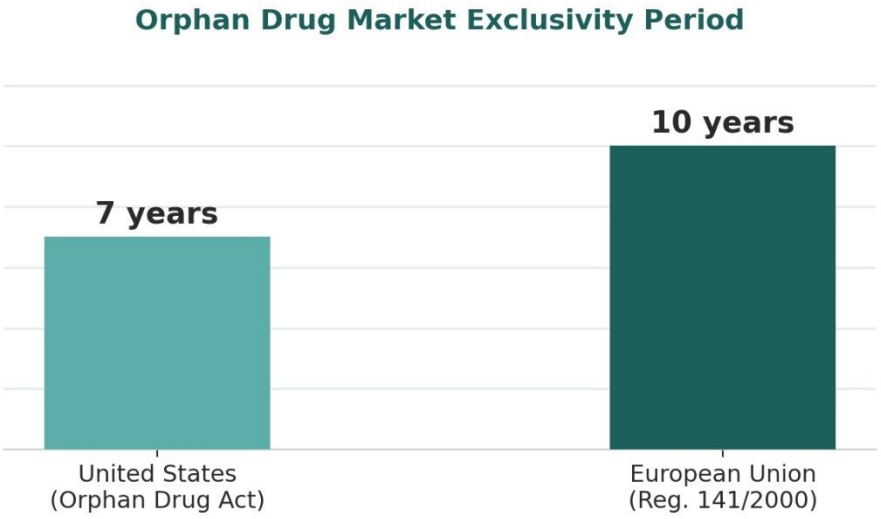

Figure 3: Orphan Drug Market Exclusivity — US vs EU

Source: U.S. Congressional Research Service; EU Regulation (EC) No. 141/2000

[18]Commonwealth Fund, 'Revisiting the Orphan Drug Act', 2025, commonwealthfund.org.
[19]Commonwealth Fund, 'Revisiting the Orphan Drug Act', 2025, commonwealthfund.org.
[20]Health Affairs, 'Evaluating the Impact of the Orphan Drug Act's Seven-Year Market Exclusivity Period', 2018.
[21]European Parliament and Council, Regulation (EC) No. 141/2000 on Orphan Medicinal Products, EUR-Lex.
[22]European Commission, Report on the Experience Acquired as a Result of the Application of Regulation (EC) No. 141/2000, COM(2008) 679, EUR-Lex.
[23]EURORDIS-Rare Diseases Europe, 'Orphan Medicines Regulation', eurordis.org.
[24]EURORDIS-Rare Diseases Europe, 'Orphan Medicines Regulation', eurordis.org.

3. United Kingdom — Differentiated Health Technology Assessment

Recognising that standard cost-effectiveness thresholds systematically disadvantage ultra-rare conditions, NICE operates a dedicated Highly Specialised Technologies (HST) programme alongside its standard technology appraisal route.[25] HST evaluations apply a substantially higher cost-effectiveness ceiling reported in recent cases at up to roughly £300,000 per quality-adjusted life year compared with the standard £20,000–£30,000 threshold, reflecting greater weight given to disease severity and the acceptance of higher evidentiary uncertainty for very small patient populations.[26] The UK has also used the Innovative Medicines Fund and outcome-based, risk-sharing commercial agreements , for example, tying continued payment for a gene therapy to real-world clinical outcomes over a multi-year period to manage the fiscal risk of extremely high one-time treatment costs.[27] Genomics England's national sequencing initiatives have separately strengthened the diagnostic side of the ecosystem.[28]

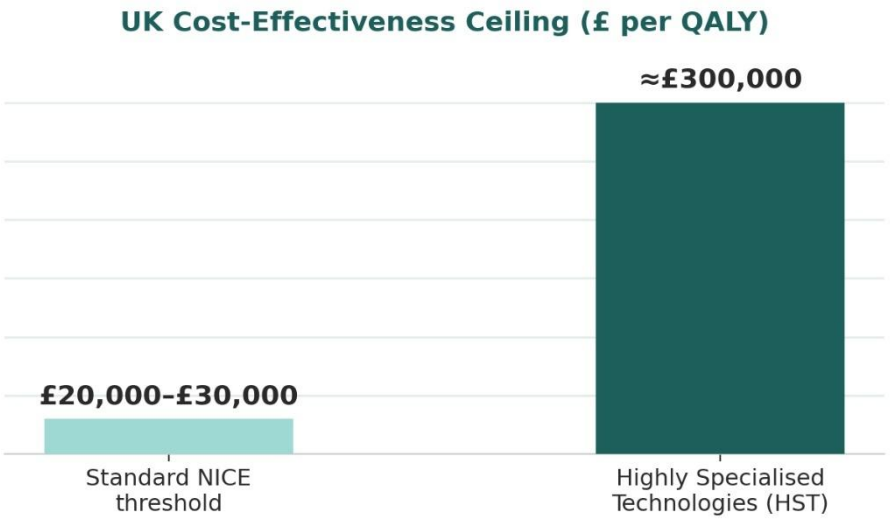

Figure 4: UK NICE Cost-Effectiveness Ceiling — Standard vs HST

Source: Health Economics Review, 2024; NICE HST programme literature

[25]National Institute for Health and Care Excellence (NICE) Highly Specialised Technologies programme literature, as reviewed in PMC10043140 and related health-policy journals.
[26]Health Economics Review, 'Comparative Policy Analysis of National Rare Disease Funding Policies in Australia, Singapore, South Korea, the United Kingdom and the United States: A Scoping Review', 2024.
[27]Health Economics Review, 'Comparative Policy Analysis of National Rare Disease Funding Policies in Australia, Singapore, South Korea, the United Kingdom and the United States: A Scoping Review', 2024.
[28]UK Parliamentary Office of Science and Technology (POST), 'Treating Rare Diseases: The Challenge of Orphan Drugs', 2025.

4. Japan — Fast-Track Approval With National Reimbursement

Japan's orphan drug system, administered by the Pharmaceuticals and Medical Devices Agency (PMDA), grants orphan designation to products for conditions affecting a small, defined patient population and offers priority review, R&D subsidies, and tax incentives, alongside near-automatic inclusion of approved orphan drugs in the National Health Insurance reimbursement list reducing the separate, often protracted, reimbursement negotiation that many other systems require. This tight coupling of regulatory approval and reimbursement is a distinguishing feature relative to jurisdictions like the UK or India, where approval and funding are decided by separate bodies on separate timelines.

5. Australia — Program-Based Funding With Risk-Sharing

Australia funds the highest-cost rare disease therapies through its Life Saving Drugs Program (LSDP), a mechanism separate from the standard Pharmaceutical Benefits Scheme, combined with confidential risk-sharing agreements between government and manufacturers that link continued funding to agreed clinical or financial conditions.[29][30] This program-based approach allows Australia to fund a small number of extremely high-cost therapies without destabilising the broader pharmaceutical benefits budget.

6. Singapore — A Resource-Constrained Comparator

Singapore is a particularly instructive comparator for India because it faces similar resource-allocation constraints without the scale of the US or EU markets. Its Rare Disease Fund matches public donations with government contributions to help finance access to high-cost therapies, while national genomics and precision-medicine initiatives aim to shorten the diagnostic pathway.[31] This blended public–philanthropic financing model offers a template that may be more readily transplantable to India's context than fully tax-funded European systems.

[29]Health Economics Review, 'Comparative Policy Analysis of National Rare Disease Funding Policies in Australia, Singapore, South Korea, the United Kingdom and the United States: A Scoping Review', 2024.
[30]BMC Health Services Research, 'The Value-for-Money Assessment and Funding Arrangements for High-Priced Drugs: A Comparative Analysis of HTA Agencies in South Korea, England, Australia, and Canada', 2025.
[31]Health Economics Review, 'Comparative Policy Analysis of National Rare Disease Funding Policies in Australia, Singapore, South Korea, the United Kingdom and the United States: A Scoping Review', 2024.

Comparative Analysis

The table below compares India's current rare disease framework with six mature rare disease ecosystems across key policy domains.

Domain	India	United States	European Union	United Kingdom	Japan	Australia
Rare disease definition	No statutory definition; policy-based recognition	<200,000 patients nationally*	≤5 in 10,000 people	≤5 in 10,000 people	Generally <50,000 patients	≤5 in 10,000 people
Orphan drug designation	No dedicated orphan designation pathway	FDA orphan designation	EMA centralised orphan designation	MHRA orphan designation	PMDA orphan designation	TGA orphan designation
Core incentive	Patient financial assistance under NPRD; no orphan-specific market exclusivity	7-year market exclusivity, tax credit, fee waivers	10-year market exclusivity, fee reductions, protocol assistance	Highly Specialised Technologies (HST) pathway and Innovative Medicines Fund	Priority review, subsidies, and 10-year re-examination period	Life Saving Drugs Program, fee waivers, and priority review
Patient financial support	Up to ₹50 lakh under NPRD	Insurance-based coverage and patient assistance programmes	Member State reimbursement mechanisms	NHS funding and Innovative Medicines Fund	National Health Insurance	Life Saving Drugs Program
National registry	National rare disease registry under development (ICMR)	Multiple disease-specific registries	European Reference Networks (ERN)-linked registries	National Congenital Anomaly and Rare Disease Registration Service (NCARDRS)	Disease-specific national registries	State and national rare disease registries
Centres of excellence	15 designated Centres of Excellence	NIH-supported specialist centres	European Reference Networks	NHS specialist centres	Designated specialist hospitals	Specialist referral centres
Newborn screening	Limited; implemented by selected states	Universal; screening panel varies by state	Varies across Member States	Universal; expanding screening panel	Universal	Universal; screening panel varies by jurisdiction

Two key patterns emerge. First all mature rare disease ecosystems combine regulatory incentives such as orphan drug designation, market exclusivity, or priority review with dedicated funding or reimbursement mechanisms designed for small patient populations. While India has introduced broader manufacturing incentives through the PLI scheme, it does

not yet have a dedicated orphan drug incentive and reimbursement framework. Second, in comparator countries, national patient registries and centres of excellence are closely linked to funding and reimbursement decisions. In contrast, India's registry and Centre of Excellence network currently operate largely independently of financing decisions.

Policy Gaps in India

Gap 1 — No Comprehensive Orphan Drug Regulatory Track
India has provisions for accelerated approval but does not have a separate regulatory pathway for orphan drugs with clear eligibility criteria and incentives, unlike the US, EU, and Japan.

Gap 2 — Limited Reimbursement Framework
The NPRD provides financial assistance of up to ₹50 lakh, but India still lacks a structured reimbursement system for high-cost rare disease treatments. PM-JAY also does not routinely cover these therapies, leaving many patients without adequate financial support.

Gap 3 — Fragmented Patient Journey
Screening, diagnosis, CoE referral, financial assistance applications, and treatment are managed by different agencies with limited interoperability, leaving patients and families to navigate the system largely unassisted.

Gap 4 — Weak Genomic and Diagnostic Integration
Genetic testing capacity is concentrated in a few metros and is not systematically linked to the CoE network or to newborn and antenatal screening programmes.

Gap 5 — Insufficient Newborn Screening
Unlike the near-universal newborn screening programmes in the US, EU, Japan, and Australia, India's coverage remains limited and largely state-dependent, delaying diagnosis for treatable Group 1 and Group 2 conditions where early intervention matters most.

Gap 6 — Underdeveloped Real-World Evidence Ecosystem
The absence of a fully operational national registry limits India's ability to generate the natural-history and real-world evidence needed for both regulatory decisions and future risk-sharing agreements with manufacturers.

Gap 7 — Limited Domestic Innovation Incentives
Beyond the general-purpose PLI Scheme, India lacks orphan-drug-specific fiscal incentives such as dedicated tax credits for orphan clinical trials that have been central to the US and Japanese models.

Gap 8 — Limited Patient Organisation Engagement in Policy Design

While patient organisations such as the Organization for Rare Diseases India and the Indian Organization for Rare Diseases actively advocate for implementation, their formal role in policy design, CoE governance, and registry oversight remains limited compared with the structured role EURORDIS plays in EU policymaking.

Policy Recommendations

Pillar 1 — Enable Earlier Diagnosis

- Introduce phased, universal newborn screening for a defined panel of treatable Group 1 and Group 2 conditions, beginning with states that already have screening infrastructure for metabolic disorders.
- Build a national genetic-testing network by accrediting regional laboratories and linking them to CoEs via telepathology and sample-referral pathways.

Pillar 2 — Improve Sustainable Access

- Introduce managed access and risk-sharing arrangements for high-cost Group 3 therapies, drawing on approaches used in the UK and Australia.
- Expand Ayushman Bharat/PM-JAY to include defined treatment packages for rare diseases, reducing reliance on one-time financial assistance under the NPRD.
- Ensure timely and dedicated disbursement of the ₹50 lakh financial assistance under the NPRD to address delays reported by patient groups.

Pillar 3 — Strengthen Research and Evidence

- Complete and mandate participation in the ICMR National Rare Disease Registry across all CoEs, with interoperable data standards.
- Fund India-specific natural-history studies for the most prevalent rare diseases on the NPRD list.
- Establish a national rare disease biobank to support future gene-therapy and genomics research.

Pillar 4 — Encourage Domestic Innovation

- Introduce orphan-drug-specific incentives — tax credits for qualifying clinical trials, accelerated CDSCO review timelines, and application-fee waivers — alongside the existing PLI Scheme.
- Support local biosimilar and biologic manufacturing for high-volume Group 2/3 therapies to reduce import dependence.

Pillar 5 — Build Patient-Centric, Decentralised Care

- Expand the CoE network toward at least one designated centre per state over the next five years, formalising referral pathways from district hospitals.

- Scale telemedicine and genetic-counselling services to reduce the burden of travel to metro-based CoEs.
- Create dedicated patient support roles at CoEs to guide families through diagnosis, treatment, and access to financial assistance.

Pillar 6 — Strengthen Governance

- Establish a standing inter-ministerial National Rare Disease Coordination Body spanning the Ministry of Health and Family Welfare, ICMR, CDSCO, and the Department of Pharmaceuticals.
- Formalise structured public-private partnerships with industry, academia, and patient organisations, giving patient groups a defined role in registry governance and CoE oversight.

Conclusion

India has established a foundational policy framework for rare diseases, but achieving meaningful patient outcomes will require a shift from isolated interventions to an integrated ecosystem that combines early diagnosis, sustainable financing, innovation incentives, robust data systems, and coordinated care. By adapting global best practices to India's healthcare context a dedicated orphan-drug track, differentiated reimbursement mechanisms, universal newborn screening and a genuinely national registry and centre network the country can improve access for patients while fostering long-term research and pharmaceutical innovation. The NPRD's five-year record shows both the value of a national policy framework and the cost of uneven implementation.

***

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