

Malaysia's Evolving Rare Disease Ecosystem and the Future of Orphan Drug Access

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Malaysia's rare disease landscape is entering a new phase, driven by the launch of its first National Policy for Rare Diseases and growing investments in diagnostics, genomics and treatment access. While the policy provides a stronger framework for coordinated care, significant gaps remain in early diagnosis, patient identification, specialist capacity, registries and sustainable reimbursement for high-cost orphan therapies. Over the next decade, stronger implementation, regional ASEAN collaboration and precision medicine could help Malaysia build a more integrated and equitable rare disease ecosystem.



Rare diseases/orphan diseases affect a very small fraction of the population and are frequently debilitating. World Health Organization (WHO) defines a rare disease as a condition that affects 1 in 2,000 of the population. Globally, there are more than 7,000 rare diseases affecting more than 300 million individuals. Approximately 80 per cent have a predominantly genetic cause. A significant unmet need exists, with around 95 per cent of rare diseases lacking an approved treatment protocol and being managed primarily symptomatically.

Malaysia, with a population of approximately 34.2 million, strengthened its rare disease landscape through the launch of its first National Policy for Rare Diseases (NPRD) on August 27, 2025, thereby establishing the country's inaugural policy framework for rare disease management. NPDR defines rare disease as a disease that affects 1 in 4000 of the population.

In Malaysia, access to orphan drugs is challenging due to their high cost. For instance, enzyme replacement therapy (ERT) for selected rare diseases costs RM 500,000–1 million annually. Orphan drugs often require out-of-pocket spending or ad hoc

government support. As a result, rare diseases are becoming a healthcare financing challenge rather than just clinical issues, requiring the universal health coverage framework to prioritise, support, and pay for high-cost, low-volume orphan drugs.

Malaysia is addressing key challenges with the NPRD policy to enable effective management of rare diseases. Key challenges include the absence of a national registry, delayed diagnosis due to limited genetic expertise and testing infrastructure, and restricted access to costly orphan drugs. These gaps create significant clinical, financial, and emotional burdens for patients and caregivers.

Recent Developments

The NPRD is strategically important for Malaysia's rare disease ecosystem as it establishes an emerging operating model by linking diagnosis, clinical management, treatment access, laboratory services, advocacy and health education, registry development, patient support, and health technology assessment into a more coordinated access pathway. This will enable Malaysia to establish a fair and sustainable healthcare system for rare disease management while moving closer to universal health coverage goals. Malaysia is participating in ASEAN-led discussions on rare disease policy and regional collaboration. These initiatives could help overcome scale-related challenges, including small patient populations, limited specialist expertise, and the high cost of orphan drugs, by enabling coordinated national-level procurement, which is otherwise difficult to sustain at the individual country level.

Apart from policy advancements, Malaysia is establishing itself as a regional hub for genomic innovation in rare diseases. This is reflected by its participation in the Human Genome Project 2 Rare Disease Alliance of the Asia-Pacific Region (HGP2 RaDiAnce-APAC) event in Kuala Lumpur in May 2026. BGI Genomics, as a technology partner, offers innovative sequencing and bioinformatics to support patient stratification, disease gene identification, and development of personalised medicines for ultra-rare diseases. The objectives of this coalition are aligned with Malaysia's NPRD, particularly its focus on strengthening laboratory services, registry development, and precision medicine.

Malaysia's emphasis on improving access to orphan therapies was further demonstrated by the regulatory approval of AstraZeneca's Soliris (eculizumab), a targeted complement inhibitor used in the treatment of several rare neurological and haematological disorders. This approval signals growing opportunities for pharmaceutical companies to enter niche, high-value markets like Malaysia. From a pharma strategy perspective, Malaysia has the potential to become a niche market for high-value orphan drugs. However, Malaysia's ability to leverage these opportunities will depend on better patient identification, local evidence generation, more transparent reimbursement processes, and sustainable funding mechanisms that balance innovation and affordability.

Malaysia has been focusing on early diagnosis through newborn screening for decades. However, expanded newborn screening for rare diseases remains limited and could support earlier diagnosis and timely intervention for patients with rare diseases, if implemented strategically.

Future Outlook

The rare disease landscape in Malaysia is expected to transition from a fragmented system to a more coordinated approach through policy development and implementation over the next 5-10 years. While the national policy provides a clear framework for rare disease management, persistent challenges such as delayed diagnosis, data gaps, high healthcare costs, and funding limitations could affect the effective implementation unless efforts are accelerated toward comprehensive rare disease management.

Malaysia should act on emerging opportunities that can potentially transform its rare disease ecosystem. The first opportunity is likely to emerge in diagnostics, especially through setting up a national rare disease registry, expanding access to genetic diagnostics, and expanding newborn screening for selected rare diseases in infants, integrated with routine screening panels, as early diagnosis can enable timely therapeutic intervention. These efforts can shorten the diagnostic pathway, improve identification of patients eligible for targeted therapies or clinical trials and support real-world data generation for regulatory and reimbursement decision-making.

Access to high-cost orphan drugs is another major opportunity, which will depend on Malaysia's ability to develop a sustainable reimbursement model, including a dedicated rare disease fund, supported through government-industry collaboration. This will reduce the burden of out-of-pocket payments and ad hoc funding while giving pharmaceutical companies clearer signals on evidence expectations, access pathways, and launch potential.

There should be an emphasis on increasing the number of rare disease specialists through targeted training for clinical geneticists, genetic counsellors, metabolic physicians, and laboratory professionals. Also, ASEAN collaboration will act as a key enabler in Malaysia's rare disease strategy. Shared registries, pooled procurement of selected orphan medicines, shared

clinical expertise, and cross-border referral pathways will help improve diagnosis, evidence generation and access to treatment in the coming years.

Moving ahead, a key challenge will be to balance access to innovative treatments and affordability, while ensuring healthcare funding remains sustainable over the long term. By implementing policies in phases, offering evidence-based reimbursement and strengthening regional collaboration, Malaysia could solidify its position as a leading ASEAN market for rare disease diagnosis, treatment access and coordinated care.

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