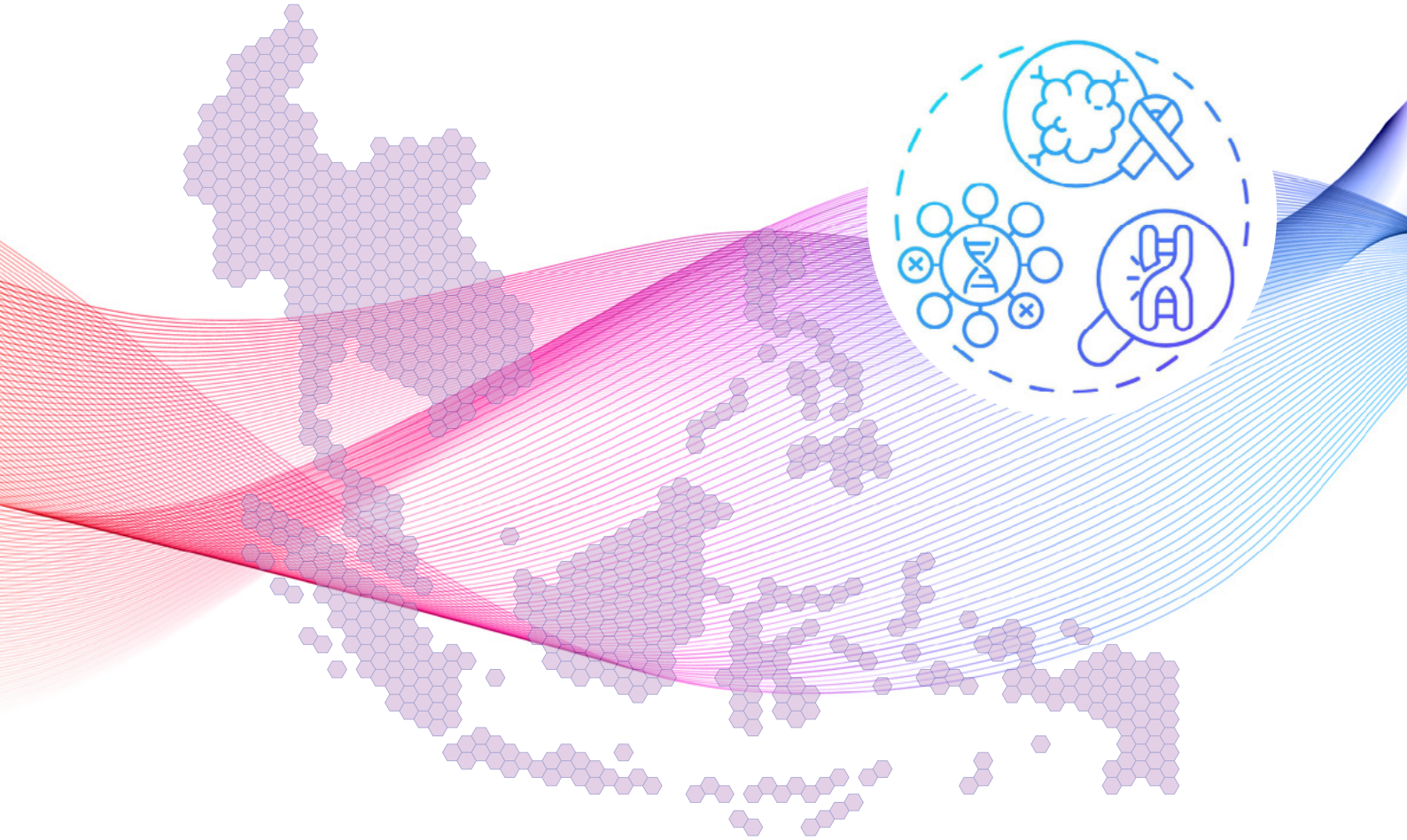
#United4Rare

ADVANCING REGIONAL COLLABORATION TO IMPROVE THE LIVES OF PEOPLE WITH RARE CONDITIONS IN THE ASEAN REGION

SOUTHEAST ASIA RARE DISEASE FORUM REPORT



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Foreword

It is my privilege to present the Report of the Southeast Asia Rare Disease Policy Forum. For too long, families across our region have navigated a fragmented landscape of diagnostic delays and limited access. In Malaysia, we believe that rarity is not a reason to deny care; in fact, it is the ultimate test of our health equity. The adoption of the World Health Assembly Resolution on Rare Diseases in May 2025 was a historic milestone. Malaysia is proud to have supported this global call, urging all nations to integrate rare diseases into their national health strategies to ensure that no patient is left behind. As I often say, “No life is too small to matter.” Nationally, our commitment is firm. With the launch of our National Policy for Rare Diseases in 2025, we have established nine strategic pillars to move beyond “ad-hoc” solutions toward a structured framework of clinical governance, precision diagnostics, and expanded treatment access. We are bringing rare conditions out of the periphery and into the heart of our mainstream health system.

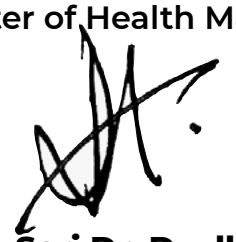
However, we cannot act alone. As Malaysia holds the ASEAN Chair, we are committed to driving this agenda across the region. The Southeast Asia Rare Disease Policy Forum, held in Putrajaya on 7 and 8 November 2025, provided the essential platform for governments, experts, and advocates to outline a shared vision for collaboration. These outcomes will be presented at the ASEAN Health Ministers Meeting (AHMM) 2026. Our goal is to guide the development of a coherent ASEAN Rare Disease Declaration and Action Plan, ensuring that our collective health systems are resilient enough to care for the 45 million individuals living with rare conditions in Southeast Asia.

I extend my sincere appreciation to Asia Pacific Alliance of Rare Disease Organisations (APARDO), Rare Diseases International (RDI), AstraZeneca, Chiesi, our technical partners, and all forum participants for their dedication. The

insights in this report will help guide the development of an ASEAN Rare Disease Declaration and Action Plan, which will support our shared vision of advancing regional collaboration to improve the lives of people with rare conditions in the ASEAN Region.

To the patients and their families—our "little heroes"—this report is for you. We remain steadfast in our whole-of-nation effort to ensure that every life, no matter how rare, is valued. Because every life is precious.

Minister of Health Malaysia



Datuk Seri Dr. Dzulkefly bin Ahmad

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Executive Summary

Rare diseases affect an estimated **45 million people** in Southeast Asia, posing a significant, persistent challenge for equity and health systems. Although each rare disease is individually uncommon, more than 7,000 conditions have been identified globally. Approximately **70-80%** are genetic in origin, and nearly **70%** present during childhood. Families across the region continue to experience long diagnostic delays, fragmented referral pathways, limited specialist capacity, and high treatment costs. These barriers result in substantial medical, social, and economic burdens.

Global recognition of rare diseases as a public health **priority** has increased considerably in recent years. The **2021 United Nations Resolution**, the **2025 World Health Organization (WHO) Executive Board recommendation**, and the **unanimous adoption of World Health Assembly (WHA) Resolution 78.11** in **May 2025** collectively mark a turning point. WHA Resolution 78.11 calls for Member States to integrate rare diseases into national health strategies, strengthen diagnostics and registries, expand specialist services, and improve equitable access to treatment. It also mandates the WHO to develop a ten-year GAPRD that will provide a tangible framework for action and technical and normative guidance for countries.

Within this evolving global context, countries within the Association of Southeast Asian Nations (ASEAN) region are advancing regionally grounded approaches that can help shape the forthcoming GAPRD. This positions ASEAN region not only as an implementer of global commitments, but as a contributor to global norm-setting informed by regional realities.

This report analyses the rare disease landscape across **six ASEAN Member States** – Indonesia, Malaysia, the Philippines, Singapore, Thailand, and Viet Nam. It draws on literature, national data, key informant interviews, and regional dialogues held during the **Southeast Asia Rare Diseases Policy Forum in Putrajaya, Malaysia, on 7-8 November 2025**. The Forum brought together more than 120 participants from governments, patient organisations, academia, research institutions, philanthropy, and industry.

Beyond documenting existing gaps and variations, the report demonstrates how ASEAN region is **progressively shaping a more coordinated approach to rare disease management through shared priorities, collective learning, and emerging regional solutions.** The insights generated support future ASEAN cooperation and contribute to early thinking on a potential **ASEAN Rare Disease Declaration and Action Plan**. Taken together, these efforts illustrate what a region can achieve when Member States act collectively to advance rare disease policy and systems strengthening.

Background and Objectives

Globally, rare diseases affect more than 300 million people, with approximately **30%** of affected children not surviving beyond the age of five. In the Asia-Pacific region, patients often endure diagnostic journeys lasting five to seven years, characterised by repeated referrals, misdiagnoses, and limited access to specialised tests. Families frequently face financial hardship due to the cost of diagnostics, specialised nutrition, travel, and long-term treatment.

Several ASEAN Member States have made notable progress. The Philippines enacted **Republic Act 10747 (2016)**, the first rare disease law in ASEAN. Malaysia launched its first **National Policy for Rare Diseases** in 2025, outlining nine strategic pillars to strengthen the system. Thailand integrates rare diseases within its Universal Coverage Scheme and includes selected orphan medicines in its **National List of Essential Medicines (NLEM)**. Singapore supports access through the **Rare Disease Fund (RDF)** and has maintained a regulatory pathway for orphan medicines since 1991. Indonesia and Viet Nam are undertaking systemic reforms to expand screening, genomic capacity, access to treatment, and regulatory processes.

Against this backdrop, the report has three main objectives:

1. To provide an overview of the current state of rare disease management in six ASEAN countries across diagnostics, referral systems, registries, treatment access, and governance.
2. To synthesise forum discussions and evidence to produce short-term, actionable recommendations that support national reforms and future ASEAN collaboration.
3. To reflect on how regional experiences within ASEAN may offer insights relevant to broader global discussions on rare disease policy and systems strengthening.

Methodology

The report uses a mixed-methods approach consisting of:

- Desk research by reviewing publicly available literature.
- Key informant interviews with policymakers, clinicians, researchers, and patient organisations.
- Deliberations from key stakeholders of the ecosystem at the **Southeast Asia Rare Diseases Policy Forum on 7-8 November 2025 in Putrajaya, Malaysia**.

Findings are presented according to four thematic areas aiming to inform the WHA Resolution Implementation and the forthcoming **GAPRD:**

1. Global Commitments and ASEAN Collaboration
2. Patient Journey from Diagnostics to Care
3. Access Pathways to Treatment
4. Innovative and Sustainable Financing

The report acknowledges key limitations, including variable definitions across of rare diseases, incomplete registries, and uneven access to treatment across countries.

ASEAN Leadership in Advancing Regional Collaboration on Rare Diseases

ASEAN's approach to emerging and complex health challenges is rooted in the principle of **ASEAN Centrality** and the vision of an **ASEAN Community** that is people-centred, inclusive, and resilient. Established in 2015, the ASEAN Community is anchored in three mutually reinforcing pillars: the ASEAN Political and Security Community, the ASEAN Economic Community, and the ASEAN Socio-Cultural Community (**ASCC**). Within this architecture, **ASCC** provides a strong foundation for addressing long-term health system challenges, including rare diseases, through its focus on human development, social well-being, universal health coverage, and the protection of vulnerable populations.

Health cooperation under the ASCC is led by the ASEAN Health Sector, organised around four priority areas (known as the **ASEAN Health Clusters**) under the ASEAN Post-2015 Health Development Agenda (APHDA): ASEAN Health Cluster 1 on Promoting Healthy Lifestyles; ASEAN Health Cluster 2 on Responding to All Hazards and Emerging Threats; ASEAN Health Cluster 3 on Strengthening Health Systems and Access to Care; and ASEAN Health Cluster 4 on Ensuring Food Safety.

Among these, **Health Cluster 3**, which particularly focuses on strengthening health systems and access to care, provides a natural basis for future rare-disease cooperation through its focus on equity, innovation, and people-centred health services. **Health Cluster 1 also provides relevant opportunities, as it focuses on** health promotion, community engagement, and early detection and aligns with efforts to raise awareness of rare diseases and improve timely care-seeking. These tools demonstrate ASEAN's capacity to translate its commitments into coordinated, multisectoral mechanisms.

As ASEAN develops its Post-2025 Vision and future health agendas, there is a strategic opportunity to mainstream rare diseases within existing regional priorities, leverage digital and regulatory cooperation, and advance more coordinated approaches to diagnosis and treatment access. Doing so would reinforce ASEAN centrality and regional solidarity while advancing more equitable and sustainable responses for people living with rare diseases.

Common barriers across ASEAN, which include:

- Limited integration of rare diseases within ASEAN's formal health cooperation mechanisms, including health sector governance structures, priority health clusters, technical working groups, and ministerial platforms that shape regional policy dialogue, coordination, and implementation.
- Limited integration of patient, family, and community perspectives in policy design and decision-making, with PLWRD and patient advocacy organisations not consistently engaged from the outset in the co-design, implementation, and evaluation of policies and programmes.
- Insufficient screening coverage and uneven access to advanced diagnostics.
- Unequal treatment availability across countries and within countries.
- High financial burden due to costly therapies and a lack of a sustainable financing mechanism.
- Fragmented governance, multisectoral coordination, and data-sharing.

Key Opportunities for Regional Action include:

- Embedding rare diseases within ASEAN health clusters and workplans.
- Harmonising diagnostics and genomic standards, in particular, newborn screening.
- Strengthening referral pathways and establishing centres of excellence.
- Adopting joint regulatory and HTA approaches.
- Ensuring meaningful participation of patients, families, caregivers, communities, and patient advocacy groups in policy development of rare disease management.

Patient Journey

Across Southeast Asia, the patient journey for rare diseases is marked by long diagnostic delays, fragmented referral pathways, and uneven access to specialised services. Families typically enter the health system through one of two routes: population-based newborn screening, where it exists, or symptom-led clinical presentation, which remains the dominant pathway for most rare conditions across the region.

Newborn screening (NBS) functions as a population-level triage mechanism for a limited set of conditions, enabling early identification only where coverage, panel scope, and follow-up systems are sufficiently developed. While NBS represents an important entry point in some ASEAN countries, its reach and integration into care vary considerably.

- **Indonesia** mandates congenital hypothyroidism screening, but national uptake remains low. Congenital hypothyroidism (CH) had a screening coverage of **4.37%** in 2023, and efforts to expand screening continue to progress.
- **Malaysia** provides subsidised testing but faces limited laboratory capacity and shortages of clinical geneticists.
- **The Philippines** screens for 29 conditions with approximately **92%** coverage, supported by PhilHealth, though confirmatory tests are not fully financed.
- **Singapore and Thailand** have comprehensive newborn screening systems and strong genomic capacity.
- **Viet Nam** continues to expand screening and genomic capacity, though national coverage remains below **50%**.

Symptom-based clinical diagnosis continues to be the main approach for the majority of rare diseases in ASEAN, especially in nations with restricted screening panels or low newborn screening (NBS) coverage. General paediatric or specialised services are frequently used to treat children and adults who exhibit developmental delays, seizures, metabolic crises, neuromuscular symptoms, or multisystem disease. In these situation, clinician awareness, access to suitable testing, and proficiency with referral systems are critical factors in diagnosis.

Regardless of entry point, diagnostic delays commonly arise at multiple stages of the diagnostic cascade:

- **Recognition and clinical suspicion**, particularly at primary or secondary care levels where rare diseases are infrequently encountered.
- **Referral navigation**, with services often concentrated in urban tertiary hospitals, requiring families to travel long distances.
- **Access to diagnostic testing**, including biochemical, metabolic, imaging, and functional studies, which may be unavailable or unaffordable.
- **Interpretation and counselling**, constrained by shortages of clinical geneticists, metabolic specialists, and genetic counsellors.
- **Actionability**, where even a confirmed diagnosis does not guarantee access to appropriate treatment or long-term care.

Furthermore, in all countries, the cost of care remains a major challenge, and families in rural or remote areas often need to travel far to obtain confirmatory testing. How referral systems are organised also affects how quickly patients receive a diagnosis. Several countries have established more structured **referral pathways:**

- **Indonesia** strengthened its referral system under Minister of Health Regulation No. 16 of 2024, although diagnostic delays persist due to long wait times, limited specialist availability, and financial barriers.
- **Malaysia** designates Hospital Kuala Lumpur as the national referral centre and connects hospitals through the Cluster Hospital Initiative. The Institute for Medical Research provides comprehensive diagnostic testing, particularly for Inborn Errors of Metabolism.
- **The Philippines** links newborn screening results to Newborn Screening Continuity Clinics for follow-up and long-term care.

- **Singapore** uses integrated electronic referrals through the National Electronic Health Record.
- **Thailand** identifies seven tertiary referral hospitals for rare and inherited metabolic diseases.
- **Viet Nam** implemented a major reform in January 2025, allowing patients with 62 rare or severe diseases to access care at any public hospital with **100%** insurance coverage and without referral letters.

Although these reforms increase access, improving referral efficiency will not eliminate diagnostic delays without adequate diagnostic resources and a sufficiently trained workforce.

Beyond diagnostics and referral systems, ASEAN is beginning to strengthen **genomic capacity** as a pillar of rare disease management. However, its system-wide impact remains constrained by workforce gaps, fragmented data infrastructures, and limited integration into routine clinical care. For instance:

- **Singapore and Thailand** are regional leaders, supported by large-scale genome sequencing programmes, secure data platforms, and integrated clinical genomics.
- **Malaysia** is developing a national repository through the MyGenom Project and strengthening ethics and data governance.
- **The Philippines** has a strong institutional capacity through the Philippine Genome Centre and national AMR genomic surveillance.
- **Viet Nam** has expanded sequencing capacity to 66 machines by 2022 and produced more than 40 million genetic variants through its national genome project.
- **Indonesia** is scaling genomic infrastructure through the Biomedical and Genome Science Initiative (BGSi) and through facilities of the National Research and Innovation Agency (*Badan Riset dan Inovasi Nasional,* BRIN).

Newborn screening and genomic testing each play meaningful roles within evolving diagnostic pathways, and their use should be guided by clinical context, available expertise, and system capacity. For many conditions, diagnosis is established through a combination of clinical assessment and laboratory evaluation, with genomic analysis contributing additional insight where appropriate. In more complex cases, genomic technologies may support greater diagnostic clarity when embedded within well-coordinated care

pathways. Ultimately, the value of any diagnostic modality depends not only on the technology itself, but also on the strength of referral systems, counselling services, and access to timely and appropriate treatment.

A centralised **rare disease registry** is a foundational component of effective rare disease policy, enabling countries to quantify disease burden, support early diagnosis, guide service planning, facilitate research, and inform financing and access decisions. Currently, no country has a fully comprehensive national registry for rare diseases. Data is scattered across hospitals, disease-specific programs, research initiatives, and new genomic platforms. Although some areas have relatively strong digital health systems, rare disease data often remains isolated within institutions and lacks interoperability.

- **Indonesia**: No national rare disease registry; existing data are fragmented across tertiary hospitals, research initiatives, and emerging genomic platforms such as BGSi.
- **Malaysia**: No single national registry yet; disease- and hospital-based databases exist, alongside efforts to strengthen national coordination and data governance under the rare disease policy framework.
- **The Philippines**: Patient data are mainly captured through programme-based systems linked to newborn screening and selected hospital registries, but there is no unified national rare disease registry.
- **Singapore**: Strong digital health and hospital information systems support rare disease care, but data remain institutionally held rather than consolidated into a single national rare disease registry.
- **Thailand**: Existing datasets are largely embedded within hospital networks and disease-specific programmes, with no fully integrated national rare disease registry.
- **Viet Nam**: Rare disease information is primarily dispersed across central hospitals and national research initiatives, with no comprehensive national registry currently in place.

Moreover, from the **patients and families' perspective**, they describe rare disease journeys as long, uncertain, and emotionally and financially exhausting, shaped by years of delayed diagnosis, repeated misdiagnoses, fragmented referrals, and heavy reliance on patient organisations to navigate care and sustain hope.

- **Indonesia and Viet Nam:** Patient journeys are frequently marked by prolonged diagnostic delays, limited access to specialists outside major cities, and substantial financial burden, with emerging patient groups playing an important role in care navigation and peer support.
- **Malaysia, the Philippines, and Thailand:** Patients benefit from expanding screening and specialised services, but continue to face referral delays, gaps in confirmatory testing and long-term care, and coordination challenges, with advocacy organisations providing critical support, awareness, and continuity of care.
- **Singapore:** More structured diagnostic and care pathways are in place for selected conditions.

Pathways to Treatment

Moving from diagnosis to treatment remains one of the most difficult stages of the rare disease journey. This challenge reflects not only the availability of medicines, but also the complex, multi-component nature of treatment for rare diseases, which extends well beyond drug provision alone. The following country snapshots highlight how treatment pathways are currently structured, including the scope of available therapies:

- **Indonesia** remains at an early stage, with treatment access largely limited to a small number of hospitals and reliant on import permits or compassionate programmes.
- **Malaysia** provides routine enzyme replacement therapy for several lysosomal storage disorders and has established clinical protocols within tertiary hospitals. In addition, NPRA currently lists more than twenty approved orphan medicines for rare diseases across biologics and new chemical entities, covering neurological, metabolic, haematological and other rare conditions.[1]
- **The Philippines** offers several Enzyme Replacement Therapies (ERTs) and orphan drugs in tertiary hospitals, with access to unregistered therapies through Compassionate Special Permits.
- **Singapore and Thailand** have the most structured access pathways. Singapore uses the Special Access Route for unregistered medicines and maintains advanced metabolic services. Thailand has added 14 orphan medicines to the National List of Essential Medicines (NLEM) and registers nearly **50%** of globally approved orphan drugs.

1 NPRA. *List of Approved Orphan Medicines (updated March 2025)*. National Pharmaceutical Regulatory Agency, Malaysia. Link

- **Viet Nam** is gradually expanding availability through new regulatory reforms and the establishment of centralised paediatric hospitals.

Financing Rare Diseases

Financing rare disease care remains a major challenge across ASEAN, where the costs of diagnosis, specialised services, and lifelong therapies frequently exceed the design limits of public health budgets and insurance schemes. While access to treatment is often discussed in terms of medicine prices, the financial burden of rare diseases extends across the entire care pathway, including diagnostics, clinical expertise, monitoring, and long-term management. Sustainable financing for rare diseases is therefore not primarily a question of combining funding sources, but of clear responsibility, governance, and prioritisation.

Global experience shows that successful rare disease financing models share several common features:

- Public leadership: In all mature systems, the public sector retains primary responsibility for financing rare disease care, particularly for high-cost therapies.
- Defined eligibility and scope: Coverage decisions are governed by clear criteria that balance equity, clinical value, and affordability.
- Incremental innovation: New financing mechanisms are introduced gradually, often through pilots, condition-specific programmes, or conditional funding arrangements.
- Protection of patients: Effective models minimise direct financial exposure for patients and families and avoid reliance on ad hoc charitable contributions at the point of care.

Experiences from countries such as **Singapore, Thailand, Taiwan, South Korea**, and **Italy** demonstrate that innovative approaches, such as dedicated budget lines, risk-sharing agreements, or structured access programmes, can improve sustainability. However, these approaches are **highly regulated and context-specific** and cannot be assumed to transfer wholesale across settings. For ASEAN member states, this implies that:

- Financing solutions must be aligned with existing health system capacity and governance;
- Public funding can serve as an important anchor for equitable access, while private and philanthropic contributions may offer complementary support where appropriate and aligned with broader system priorities; and
- Long-term sustainability tends to be strengthened when supported by transparency, accountability, and consistent political commitment, rather than relying on informal or fragmented funding arrangements.

The combined insights indicate that regional collaboration within ASEAN is most effective in improving diagnostic outcomes when it emphasises strengthening comprehensive diagnostic pathways, rather than treating newborn screening, genomics, or registries as separate solutions. Furthermore, investment priorities have varied across Southeast Asia, leading to diverse patient and family experiences. This has also resulted in a varied national focus when developing expertise and systems that can be shared, providing an opportunity for implementation efficiencies.

Regional Priorities

ASEAN's experience highlights how regional cooperation can be structured pragmatically in contexts marked by shared constraints in capacity, financing, and system readiness. Opportunities for collaboration can be understood across three progressive and complementary tiers, reflecting differing levels of feasibility and political sensitivity among Member States.

These tiers represent optional pathways that countries may choose based on their readiness and national priorities, rather than sequential obligations.

- **Tier 1 – Sharing** focuses on the exchange of knowledge and experience, including policy models, diagnostic and referral pathways, health technology assessment approaches, and governance frameworks.
- **Tier 2 – Cooperation** builds on this through more structured engagement, such as joint workforce training, alignment of rare disease datasets and coding systems (including ORPHA and ICD), subregional diagnostic referral arrangements, and quality assurance networks.
- **Tier 3 – Collaboration** involves longer-term, selective mechanisms, including shared genomic or reference laboratory functions and regional platforms for dialogue and negotiation, where collective

approaches may improve affordability and access. *(Further details on regional priorities are provided in Chapter 8).*

Five Key Takeaways

1. Rare diseases remain a significant and under-addressed burden in Southeast Asia, with millions affected by delayed diagnosis, fragmented care and whole of life support, and high financial hardship.
2. Diagnosis is constrained by system bottlenecks, not technology gaps alone: limited screening coverage, weak referral pathways, workforce shortages, and uneven access to confirmatory testing remain the dominant barriers.
3. Treatment access is highly uneven and primarily constrained by financing, with availability often dependent on tertiary hospitals, exceptional access routes, or discretionary funding.
4. Registries and data systems are foundational enablers, necessary for planning, prioritisation, and sustainable financing, but must be designed as practical, minimum-function tools rather than aspirational platforms.
5. ASEAN can accelerate progress through differentiated, capacity-aligned collaboration, focusing first on shared learning, referral strengthening, and data foundations, while reserving deeper integration for countries that are ready.

The Southeast Asia Rare Diseases Policy Forum held in Putrajaya on 7-8 November 2025 confirmed a clear regional mandate: **ASEAN must act collectively to close persistent gaps in rare disease diagnosis, care, and financing**. The forum brought together Member States and stakeholders to define priority areas and produce recommendations that can strengthen national systems and guide coherent regional action. These measures aim to improve early detection, expand equitable access to treatment, and build sustainable, people-centred responses to rare diseases across Southeast Asia. Together, they can also form the basis for the ASEAN Rare Disease Declaration that Malaysia intends to propose.

- **Integrate Rare Diseases into ASEAN's Health Agenda and Build Long-term Regional Solidarity.** Embed rare diseases within existing

ASEAN health cooperation mechanisms to sustain regional dialogue and action.

- **Strengthen Early Detection and Diagnosis.** Prioritise improved screening, genomic capacity, and multi-stakeholder collaboration.
- **Improve Equitable Pathways to Treatment.**
- **Explore Innovative and Sustainable Financing for Rare Disease Management.**
- **Strengthen National Governance Across Member States.** Reinforce institutional coordination, accountability, and multi-sectoral support for rare-disease communities.

Recommendation

The recommendations aim to strengthen national rare disease systems while supporting practical and collaborative engagement at the regional level across ASEAN. These recommendations are designed as readiness-based, stepwise actions that recognise considerable variation in system capacity, available resources, and stage of development. Progression to subsequent actions depends on the effective establishment of essential foundational system components.

1. **Integrate Rare Diseases into ASEAN's Health Agenda and Build Long-term Regional Solidarity**

   ASEAN may consider to formally integrate rare diseases into existing regional health cooperation mechanisms to provide sustained political visibility, policy continuity, and structured regional dialogue. Integrating rare diseases into ASEAN's health agenda will create the necessary institutional foundation for coordinated action, while avoiding the imposition of uniform obligations on Member States.

2. **Strengthen Early Detection and Diagnosis**

   Member States are encouraged to prioritise strengthening early detection and diagnosis by improving end-to-end diagnostic pathways, including screening, referral systems, confirmatory testing, workforce capacity, and the responsible integration of genomics. Achieving early diagnosis requires more than just expanding technology. It is crucial to strengthen the entire diagnostic process to reduce delays and address inequities.

3. **Improve Equitable Pathways to Treatment**

   Member States are encouraged to strengthen pathways from diagnosis to treatment by clarifying eligibility, referral, and access mechanisms

for rare disease therapies within national health systems. Without equitable pathways to treatment, improvements in diagnosis may not fully translate into better treatment outcomes and could contribute to unmet needs.

4. **Explore Innovative and Sustainable Financing for Rare Disease Management**

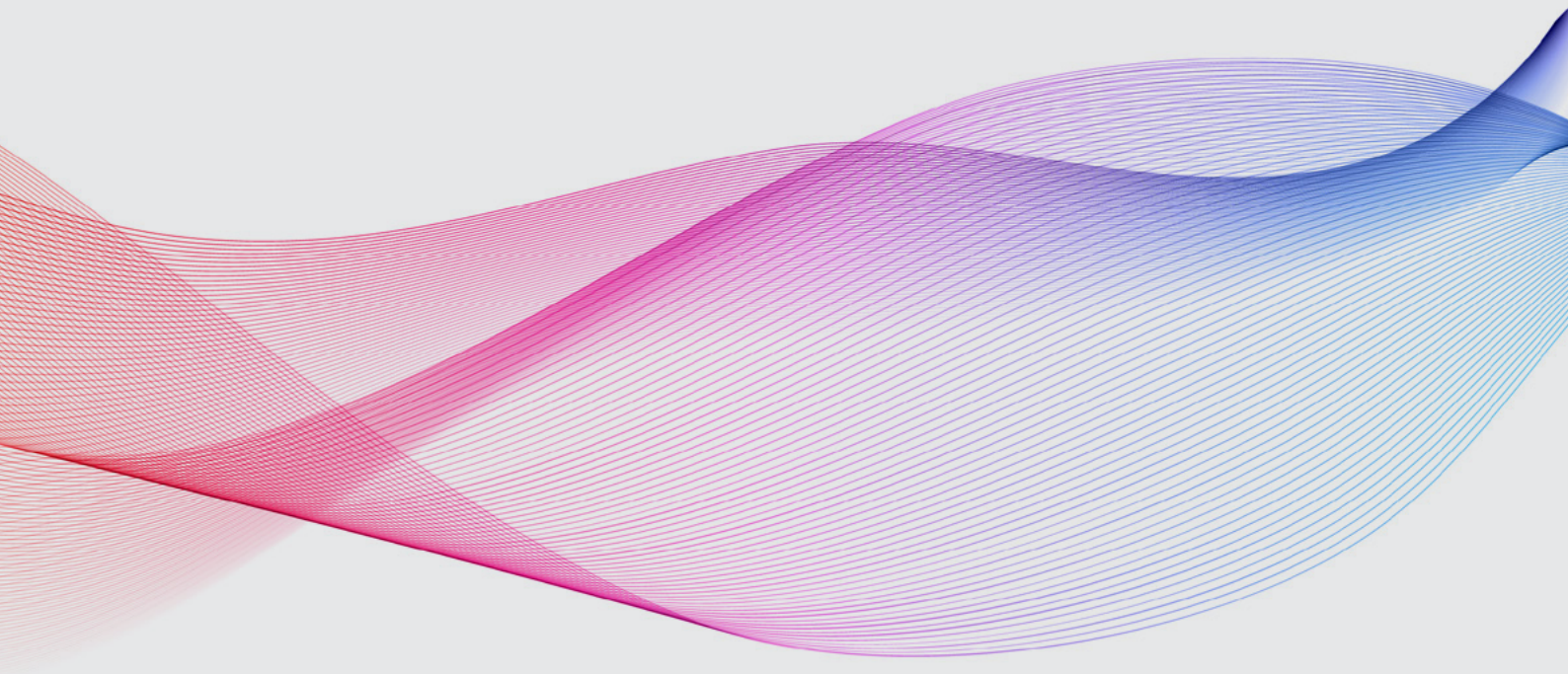
   ASEAN Member States are encouraged to explore innovative financing mechanisms for rare disease care while prioritising near-term financial protection and long-term system sustainability. Innovative financing mechanisms are viable only when supported by strong data, governance, and prioritisation frameworks.

5. **Strengthen National Governance for Rare Diseases Across Member States**

   Member States are encouraged to strengthen national governance for rare diseases by establishing clear institutional leadership within health ministries and supporting it with pragmatic, policy-relevant data systems. Effective governance, supported by practical data, is essential to enable evidence-based decision-making and accountable resource allocation for rare diseases.

CHAPTER 1.

Background and Objectives



CHAPTER 1.

Background and Objectives

Rare diseases are increasingly recognised as an important global health and equity challenge. The WHO defines a rare disease as a health condition affecting fewer than 1 in 2,000 individuals.[2] Although individually uncommon, more than 7,000 rare diseases have been identified, affecting an estimated 300 million people worldwide.[3] 70-80% are genetic in origin, and nearly 70% present during childhood. Mortality is substantial, with approximately 30% of affected children not surviving beyond the age of five.[4]

PLWRD in the Asia-Pacific region often endure long and fragmented journeys before receiving a diagnosis. On average, it takes 5-7 years before patients obtain diagnostic clarity, typically after multiple referrals and misdiagnoses.[5] Families also experience stigma, discrimination, and psychosocial distress. Financial hardship is a defining feature of the rare disease experience, driven by the high cost of diagnostic tests, specialised treatments, travel to tertiary centres, and loss of income due to caregiving responsibilities. Although treatments exist for certain conditions, effective therapies are available for only around 5% of rare diseases, and even where available, affordability remains a significant barrier without public subsidies or philanthropic support.[6]

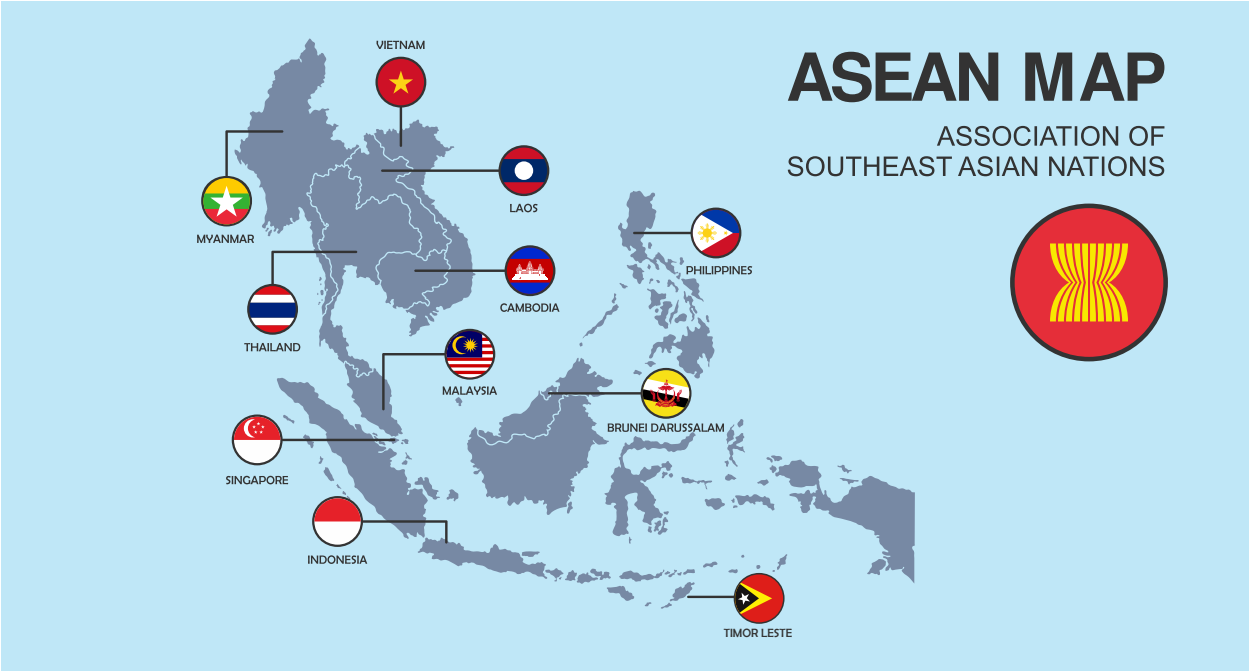
However, globally, there has been some progress. The United States Orphan Drug Act of 1983, the European Union's rare disease regulations of 2000, and similar frameworks in Japan, Taiwan, and South Korea created incentives that stimulated research and drug development, accelerated approvals, and increased availability.[7,8] Advances in genomics, particularly whole-genome

2 World Health Organization. (2025). Executive Board 156th Session – Rare diseases: a global health priority for equity and inclusion. In *who.int* (EB156/CONF./2). WHO. Link
3 The Lancet Global Health Editorial. (2024). The landscape for rare diseases in 2024. *The Lancet Global Health, 12*(3), e341. Link
4 Ibid.
5 Singh, P., Bhardwaj, R., & Subramaniam, D. (2024). Unlocking Solutions for Rare Diseases in APAC. In *iqvia.com*. IQVIA. Link
6 Ibid.
7 Shafie, A. A., Chaiyakunapruk, N., Supian, A., Lim, J., Zafra, M., & Hassali, M. a. A. (2016). State of rare disease management in Southeast Asia. *Orphanet Journal of Rare Diseases, 11*(1). Link
8 Laurini, G. S., Nikitina, V., Broccoli, M., Montanaro, N., & Motola, D. (2025). Overview of orphan Medicines in European Union: An Analysis of Regulatory and Technical–Scientific aspects. *Ther Innov Regul Sci, 59*(1087–1097). Link

sequencing (WGS), have transformed diagnostic capacity for neurological and metabolic conditions, allowing earlier and more precise identification. Importantly, the 2021 United Nations General Assembly Resolution on PLWRD and Their Families, and the 2025 WHA Resolution on Rare Diseases elevated these conditions onto the global health agenda.[9] Both frame rare diseases as matters of equity, inclusion, and universal health coverage (UHC). These developments provide momentum for countries and regions, including the ASEAN, to prioritise rare disease management as part of broader health system strengthening.

ASEAN is a regional intergovernmental organisation comprising eleven Member States of Brunei Darussalam, Cambodia, Indonesia, Lao PDR, Malaysia, Myanmar, the Philippines, Singapore, Thailand, Viet Nam, and, most recently, Timor-Leste. With a total population of more than 670 million and encompassing a wide spectrum of economic contexts, from lower-income to upper-income economies, ASEAN plays a central role in shaping regional and global cooperation across health, social development, economics, and science and technology. Its mechanisms support policy dialogue, regulatory alignment, and cross-border collaboration, while fostering shared commitments to equity and inclusive growth. Engaging ASEAN is therefore critical for strengthening rare disease responses, as the platform enables Member States to harmonise approaches, expand access to diagnostics and treatment, pool expertise, and embed rare disease priorities within broader agendas on UHC and health system resilience.

Picture 1. ASEAN Map



9 World Health Organization. (2025). Executive Board 156th Session – Rare diseases: a global health priority for equity and inclusion. In *who.int* (EB156/CONF./2). WHO. Link

Over more than five decades, ASEAN has evolved into the central convening platform for regional cooperation in Southeast Asia, shaping regional norms, mechanisms, and institutions that support dialogue, stability, and collective responses to shared challenges. This role is formally anchored in the ASEAN Charter, signed in Singapore on 20 November 2007 and entering into force in 2008, which codified ASEAN's central position in the regional architecture. The Charter then formally established ASEAN centrality as a core principle guiding the organisation's internal development and external engagement.[10]

In practice, ASEAN Centrality reflects the organisation's long-standing function as a trusted convener and facilitator of cooperation, rather than a power-based bloc. Through ASEAN-led platforms and dialogue mechanisms, Member States have cultivated habits of consultation, consensus building, and collaborative problem solving across political, economic, and socio-cultural domains. This approach has enabled ASEAN to advance cooperation on complex and sensitive issues while safeguarding inclusiveness, strategic autonomy, and regional ownership, even amid significant diversity in national contexts and capacities.[11]

The institutional expression of this cooperation is the ASEAN Community, formally established in 2015 and anchored in three mutually reinforcing pillars: the ASEAN Political-Security Community, the ASEAN Economic Community, and the ASEAN Socio-Cultural Community.[12] Together, these pillars provide a comprehensive framework for regional integration, people-centred development, and coordinated action on transboundary challenges. Within this architecture, health, social development, and the protection of vulnerable populations are core priorities, particularly under the ASEAN Socio-Cultural Community, which emphasises quality of life, inclusiveness, resilience, and people-oriented cooperation. This community-based approach provides an important foundation for addressing complex and long-term health system challenges, including rare diseases.

ASEAN today represents a relatively solid regional platform, with established institutions, shared principles, and cooperation mechanisms that can support collective action on emerging public health priorities. At the same time, the pace and depth of national progress across specific health domains are not uniform. This dynamic is particularly evident in rare disease management, where common challenges coexist with highly diverse levels of system readiness.

Progress within Southeast Asia also reflects wide variation across countries. The Philippines became the first ASEAN Member State to establish comprehensive

10 Association of Southeast Asian Nations. (2008). *The ASEAN Charter*. ASEAN Secretariat. Link
11 Anwar, D. F. (2023). ASEAN Centrality: Opportunities and Challenges. In *The Indo-Pacific Theatre: Strategic Visions and Frameworks* (pp. 155–170). Routledge. Link
12 ASEAN Secretariat. (2023). *Fact Sheet: ASEAN Community*. Link

rare disease legislation through Republic Act 10747 (2016), creating a legal foundation for diagnosis, care, and access to therapies. Malaysia has more recently advanced its National Policy for Rare Diseases (NPRD) 2025, which outlines nine strategic pillars to guide system-wide strengthening. Thailand provides UHC for selected rare diseases through coverage of diagnosis, treatments and management. Selected orphan medicines are reimbursed under the NLEM E2 category.[13,14] Singapore supports access through the Medicines (Orphan Drugs) (Exemption) Order, first introduced in 1991 and updated in 2005 and 2016, complemented by philanthropic assistance through the Rare Disease Fund. In other ASEAN Member States, including Indonesia, Viet Nam, Cambodia, Lao PDR, and Myanmar, formal rare disease frameworks are still evolving, and access to diagnostic services and treatment is shaped by a combination of existing health-system provisions, out-of-pocket spending, charitable initiatives, and compassionate-use pathways.

7,000+ conditions

- ~300 million affected globally
- ~45 million affected in SEA Region
- 70-80% genetic -> 70% begin in childhood

1.1 World Health Assembly Resolution 78.11 (2025) on Rare Diseases

Adopted on **24 May 2025**, the WHA Resolution titled *"Rare Diseases: A Global Health Priority for Equity and Inclusion"* represents a major milestone in global rare disease advocacy. Led by Egypt and Spain, and co-sponsored by **41 Member States**,[15] the Resolution highlights the need for Member States to integrate rare diseases into national health planning by developing and implementing national policies, effective programmes, and actions. This includes establishing primary and secondary evidence-based preventive measures and strategies, as well as strengthening healthcare services and financing for PLWRD.

13 Adachi, T., El-Hattab, A. W., Jain, R., Nogales Crespo, K. A., Quirland Lazo, C. I., Scarpa, M., Summar, M., & Wattanasirichaigoon, D. (2023). *Enhancing equitable access to rare disease diagnosis and treatment around the world: A review of evidence, policies, and challenges. International Journal of Environmental Research and Public Health, 20*(6), 4732. Link

14 National Health Security Office. (2024). *National Health Security Office (NHSO).* Link

15 Co-sponsoring countries: Bahrain, Bangladesh, Barbados, Bhutan, Brazil, Chile, China, Colombia, Cuba, Dominican Republic, Ecuador, Egypt, El Salvador, France, Ghana, Greece, Guatemala, Honduras, India, Iraq, Ireland, Jordan, Kuwait, Libya, Luxembourg, Malaysia, Pakistan, Palestine, Panama, Peru, Philippines, Qatar, Romania, Russia, Slovakia, Somalia, Spain, Thailand, Tunisia, Uruguay, and Vanuatu.

The preamble recognises the chronic, complex, and often disabling nature of rare diseases and acknowledges the long diagnostic odysseys and the significant social and financial burdens families face. It situates rare diseases within global commitments such as the **Sustainable Development Goals (SDGs**) and UHC.

The operative section of the Resolution urges Member States to:

- Integrate rare diseases into national health plans
- Expand newborn screening and early diagnosis
- Strengthen health systems, specialist capacity, and centres of excellence
- Establish national registries and improve epidemiological data
- Invest in research and innovation
- Involve patient organisations in policymaking processes.

At the global level, the World Health Organization is requested to develop a comprehensive ten-year GAPRD, accompanied by guidelines, data frameworks, and mechanisms to support equitable access to diagnosis, treatment, and long-term care. In line with the Resolution, the WHO is expected to submit a draft Global Action Plan on Rare Diseases for consideration by the **Executive Board at its 162nd session**, with the intention of submitting the final draft to the **Eighty-first WHA in 2028** for adoption. This process aims to strengthen international collaboration and provide technical support to countries seeking to close persistent care gaps. Taken together, the Resolution reflects a shift from fragmented, disease-by-disease responses toward a more systemic and inclusive agenda. It acknowledges both the vulnerability and the resilience of people living with rare diseases and positions their needs as integral to the UHC agenda, underscoring that addressing rare diseases is essential to building fairer and more responsive health systems worldwide.

The GAPRD is envisioned as a comprehensive roadmap that brings together equity, evidence, and accountability. Its key components include:[16]

16 World Health Organization. (2025). 78th WHA Assembly – Rare diseases: a global health priority for equity and inclusion. In *who.int* (WHA78.11). Link

- A comprehensive framework to ensure equitable, timely, cost-effective, and evidence-based diagnosis, treatment, and management of rare diseases, consistent with universal health coverage and social determinants of health.
- Strategies to strengthen data collection, research, and surveillance on rare diseases to improve understanding, enable early identification, and promote collaboration across Member States, while also encouraging greater investment in research.
- Guidelines for creating national and regional registries to support screening, monitoring, and long-term management of rare diseases.
- Global targets and strategic objectives, paired with clear accountability and monitoring mechanisms, to expand equitable access to essential diagnostics, treatments, and healthcare services for persons living with rare diseases.

During the Southeast Asia Rare Disease Policy Forum, Rare Diseases International (RDI), which represents 140 member organisations and a coalition of over 300 civil society organisations, announced that, together with its members and partners, it is establishing 6 regional task forces – one per WHO region - beginning in Asia-Pacific (the Western Pacific Region and the Southeast Asia Region) and Latin America. These task forces are intended to support the development and early implementation of the GAPRD by ensuring regional contexts are considered in the establishment of global priorities and targets, coordinating stakeholders, building capacity, and facilitating knowledge exchange. They will also help generate and share evidence and monitor progress, ensuring that efforts remain aligned and coherent across countries. The task forces are expected to serve as a regional engine for collaboration, linking patient groups and other civil society organisations in the region, and WHO regional offices to translate the Resolution into tangible action. For example, the Western Pacific Regional Task Force and the Southeast Asia Regional Task Force will link ASEAN, APARDO, and other civil society organisations in the regions as well as the respective WHO regional offices.

1.2 Rare Disease Progress in ASEAN Member States

ASEAN's existing health priorities under the ASEAN Post-2015 Health Development Agenda 2021 to 2025 provide a considerably ready platform for advancing rare disease action, enabling the region to shape global and regional agendas through its own health system experiences, rather than merely responding to external frameworks. Under the health

development agenda, regional cooperation is organised into four clusters that collectively span health promotion, emergency preparedness, health-system strengthening, and food safety. These clusters cover healthy lifestyles, communicable and emerging threats, universal access to quality health services and innovation, along with coordinated approaches to food safety and regulatory risk analysis.

The latter focuses on strengthening health systems, universal health coverage, essential medicines, human resources for health, and digital health, and provides the strongest foundation for regional rare-disease policy discussions. The leadership of Malaysia, the Philippines, and Thailand in sponsoring the WHA Resolution adds momentum to ASEAN's alignment with global commitments. This is critical, given the estimated 45 million people living with rare diseases in Southeast Asia, roughly 9% of the population. The actual burden is likely higher because surveillance is weak, registries are fragmented, and reporting is inconsistent. With around 80% of rare diseases genetically driven and nearly 70% presenting in childhood, the strain on paediatric systems and families is substantial.[17,18]

Country progress varies considerably:

- **Malaysia**: Introduced its **National Policy for Rare Diseases (2025)** with nine strategic pillars.
- **Philippines**: First ASEAN Member State to enact comprehensive rare disease legislation through **Republic Act 10747 (2016)**.
- **Singapore**: Supports access through the **Medicines (Orphan Drugs) (Exemption) Order** (1991, revised 2005 and 2016) and the Rare Disease Fund.
- **Thailand**: Provides UHC coverage for selected rare diseases through ring-fenced financing and the **NLEM E2** category for orphan medicines.
- **Indonesia, Viet Nam, Cambodia, Lao PDR, Myanmar**: No dedicated legislation; access largely depends on out-of-pocket expenditure, charitable initiatives, or compassionate-use pathways.

Several countries have scaled newborn screening panels and improved early detection pathways, while others are investing in genomic capacity and hospital-based registries. Access to rare disease treatments is also evolving, particularly through tertiary centres and public financing initiatives in some settings, encompassing a range of therapeutic approaches such as metabolic and nutritional interventions and enzyme replacement therapies; however, access remains uneven, and in many contexts continues to be constrained by limited coverage and availability.

17 Shafie, A. A., Chaiyakunapruk, N., Supian, A., Lim, J., Zafra, M., & Hassali, M. a. A. (2016). State of rare disease management in Southeast Asia. *Orphanet Journal of Rare Diseases, 11*(1). Link

18 Dumbuya, J. S., Zeng, C., Deng, L., Li, Y., Chen, X., Ahmad, B., & Lu, J. (2025). The impact of rare diseases on the quality of life in paediatric patients: current status. *Frontiers in public health, 13*, 1531583. Link

Progress on newborn screening, data systems, treatment access, and advocacy **varies widely across ASEAN**:

- Screening coverage is well established in Singapore and Thailand, more limited in the Philippines and Viet Nam, and remains at an early stage of development in Indonesia.
- While some ASEAN Member States maintain hospital-based or programme-specific lists of patients, none has a comprehensive national rare disease registry. Existing records are typically confined to tertiary hospitals, specialty clinics, or research initiatives.
- Enzyme replacement therapies are subsidised only in Malaysia and Thailand; elsewhere access depends on out-of-pocket or charitable and industry support.
- Patient Advocacy Groups (PAGs) fill critical gaps in awareness, navigation, and policy engagement. These groups are generally coordinated by the Asia Pacific Alliance of Rare Disease Organisations (APARDO).

Patient advocacy provides an essential lens for understanding regional progress, as advocacy groups often fill gaps in awareness, service navigation, and policy engagement where formal systems remain limited. Organisations such as the Philippine Society for Orphan Disorders, the Rare Disorders Society Singapore, and the Malaysian Rare Disorders Society have played central roles in shaping public discourse, supporting families, and promoting policy reform. In Malaysia, the Rare Diseases Alliance Foundation Malaysia (RDAFM) unifies multiple patient groups. It strengthens coordination across conditions, while regional networks such as Rainbow Across Borders demonstrate the growing potential for cross-country collaboration and shared advocacy within ASEAN. In Thailand, the Thai Rare Disease Foundation (ThaiRDF), established in 2016, has been supporting the development and strengthening of several disease-specific patient support groups, as well as participating in multistakeholder meetings, shaping HTA policy, and developing a new benefit package for rare diseases under UHC.[19] Furthermore, organisations such as the APARDO unites patient-advocate leaders across the Asia-Pacific to amplify rare-disease voices, strengthen shared learning, and support collaboration among disease-specific groups, national societies, and regional alliances.

ASEAN can also draw lessons from the APEC Rare Disease Action Plan of 2018,[20] which provided a structured framework through ten priority pillars spanning policy alignment, early diagnosis, research, data infrastructure, and financial support. APEC's experience highlights the need for collaboration across government, academia, and industry, the value of harmonised definitions and regulatory approaches, and the role of shared

19 Thai Rare Disease Foundation. Link
20 Asia-Pacific Economic Cooperation. (2018). *APEC Action Plan on Rare Diseases*. APEC. Link

registries in addressing small patient populations and limited expertise. It further emphasises that UHC cannot be achieved without addressing rare diseases and that coordinated regional frameworks are essential to reduce misdiagnosis, strengthen access to therapies, and promote social inclusion. These insights are relevant, as this report examines rare-disease systems in six Southeast Asian countries: the Philippines, Singapore, Malaysia, Indonesia, Viet Nam, and Thailand.

10 Pillars of the APEC Action Plan on Rare Diseases

1. Define rare diseases and orphan products with policies and processes.
2. Raise public and political awareness of rare disease issues.
3. Promote innovative research and development
4. Build human resource capacity in medical, nursing, nutrition, and other allied health and non-health sectors
5. Facilitate early, accurate, and systematic diagnosis
6. Coordinate patient-centred care across medical and other health disciplines, life course, and location.
7. Deliver new and accessible treatments to patients.
8. Support financial and social needs of patients and their families
9. Manage pooling and usage of patient data securely and effectively.
10. Prioritise comprehensive domestic rare disease policy integrating Pillars 1-9.

1.3 SEA Rare Diseases Policy Forum

> ***This forum marks a turning point – where Southeast Asia moves from awareness to action and from commitment to real, tangible care."***

YB Datuk Seri Dr. Dzulkefly Ahmad

Minister of Health Malaysia

The Southeast Asia (SEA) Rare Diseases Policy Forum was held to provide a regional platform for advancing collaboration on rare disease policy within ASEAN. The forum was chaired by **the Ministry of Health Malaysia**, in its capacity as ASEAN Chair for 2025. It was organised by **APARDO**, **in collaboration with RDI, with support from AstraZeneca and Chiesi**. The closed-door event gathered **over 120 participants** representing governments, patient advocacy organisations, clinicians, researchers, industry, and philanthropic partners. Delegations from the six focus ASEAN countries (Malaysia, Indonesia, the Philippines, Singapore, Thailand, and Viet Nam) convened to share national perspectives and engage in dialogue. The forum was held in Putrajaya, Malaysia, on 7-8 November 2025, structured around dialogues and exchanges amongst all participants in plenary panels, breakout sessions, and country roundtables.

This event:

- **Built momentum and gathered expert inputs** to support the development of an ASEAN Rare Diseases Declaration and Action Plan, under the leadership of the Ministry of Health Malaysia.
- **Shared ASEAN country experiences** on rare disease situations, challenges, and opportunities.
- **Built multi-stakeholder dialogue** across governments, clinicians, researchers, industry, and patients.
- **Explored the potential for regional cooperation** on diagnosis, innovation, care, and financing on Rare Disease.

The forum discussions centred on four main themes. The first focused on translating global commitments, including the WHA Resolution, into ASEAN strategies and national action plans through stronger multistakeholder collaboration. The second addressed the patient journey from diagnostics to care, highlighting ways to expand newborn screening, strengthen referral systems, advance genomics and digital health, and share best practices for registries and care coordination. The third emphasised equitable access to treatment by drawing on international and regional good practices and identifying strategies that ASEAN can adapt to improve the availability, affordability, and sustainability of therapies for rare diseases. The last explored innovative and sustainable financing for rare disease management. This session focused on how ASEAN governments could strengthen fiscal policies, budget mechanisms, and partnerships to secure funding for rare diseases.

Building on these discussions, Malaysia reiterated its strong commitment to elevating the forum's outcomes within ASEAN's formal health cooperation mechanisms. As ASEAN Chair for 2025 and host of the forthcoming ASEAN Health Ministers Meeting 2026, Malaysia will present the key findings and proposed areas of collaboration to Member States for their consideration and guidance. The Ministry of Health also conveyed its intention to work closely with ASEAN counterparts, the ASEAN Secretariat, and technical partners to explore the development of a regional roadmap on rare diseases that aligns with the WHA Resolution and supports ASEAN's post-2025 health agenda. This commitment underscores Malaysia's dedication to fostering solidarity, promoting inclusive dialogue, and strengthening sustainable cooperation across the region in support of people living with rare diseases.

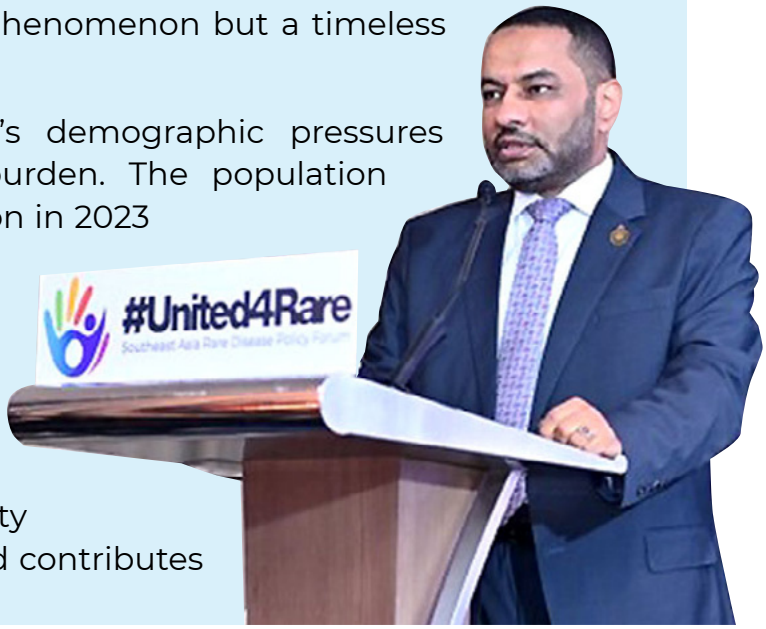
1.4 Objectives of the Report

The present report has two main objectives:

1. To provide an overview of the current state of rare disease care in ASEAN, including national priorities, policies, programmes, and systemic gaps. The analysis covers governance arrangements, financing mechanisms, service delivery models, and country-level progress in diagnostics, registries, genomics, and access to treatment.
2. To synthesise discussions and existing evidence to generate short-term, actionable recommendations, with a focus on strengthening diagnostic capacity and referral systems, developing registries and data-sharing platforms, and advancing equitable access to therapies through regulatory innovation and evidence-informed policy.

From Ancient Foundations to Modern Rare Disease Care in Egypt

In his keynote address at the Southeast Asia Rare Disease Policy Forum on 7 November 2025 in Putrajaya, Malaysia, **Prof. Mohamed Hassany** traced Egypt's long and evolving relationship with rare diseases, beginning not in the 20th century, but in antiquity. He opened with the Ebers Papyrus, a 1500 BC medical text displayed in Leipzig that contains more than 700 magical formulas and early descriptions of congenital abnormalities, neonatal assessment practices, and clinical observations. Its existence, he argued, underscores that rare diseases are not a modern phenomenon but a timeless human reality.



In the present day, Egypt's demographic pressures amplify the rare disease burden. The population reached more than 108 million in 2023 and is projected to exceed 120 million by 2030. Although fertility has declined from 2.7 to 1.9 children per woman; high birth rates remain concentrated in rural and border areas. Consanguinity reaches 18% of marriages and contributes

significantly to congenital and genetic disorders, which are among the leading causes of neonatal mortality. Furthermore, in Egypt, an estimated 590,000 people live with rare diseases, and 263 genetic disorders have been documented in national catalogues.

Egypt's structured response began in 1999 with neonatal screening for congenital hypothyroidism, expanded in 2015 to include phenylketonuria, and scaled up in 2021 through the Presidential Initiative for Early Detection of 19 Genetic Diseases. The most frequently detected disorders include G6PD deficiency, galactosemia, and cystic fibrosis. A major study from Ain Shams University found that 4% of paediatric admissions involved genetic diseases, primarily neurological, hematologic, and chromosomal conditions.

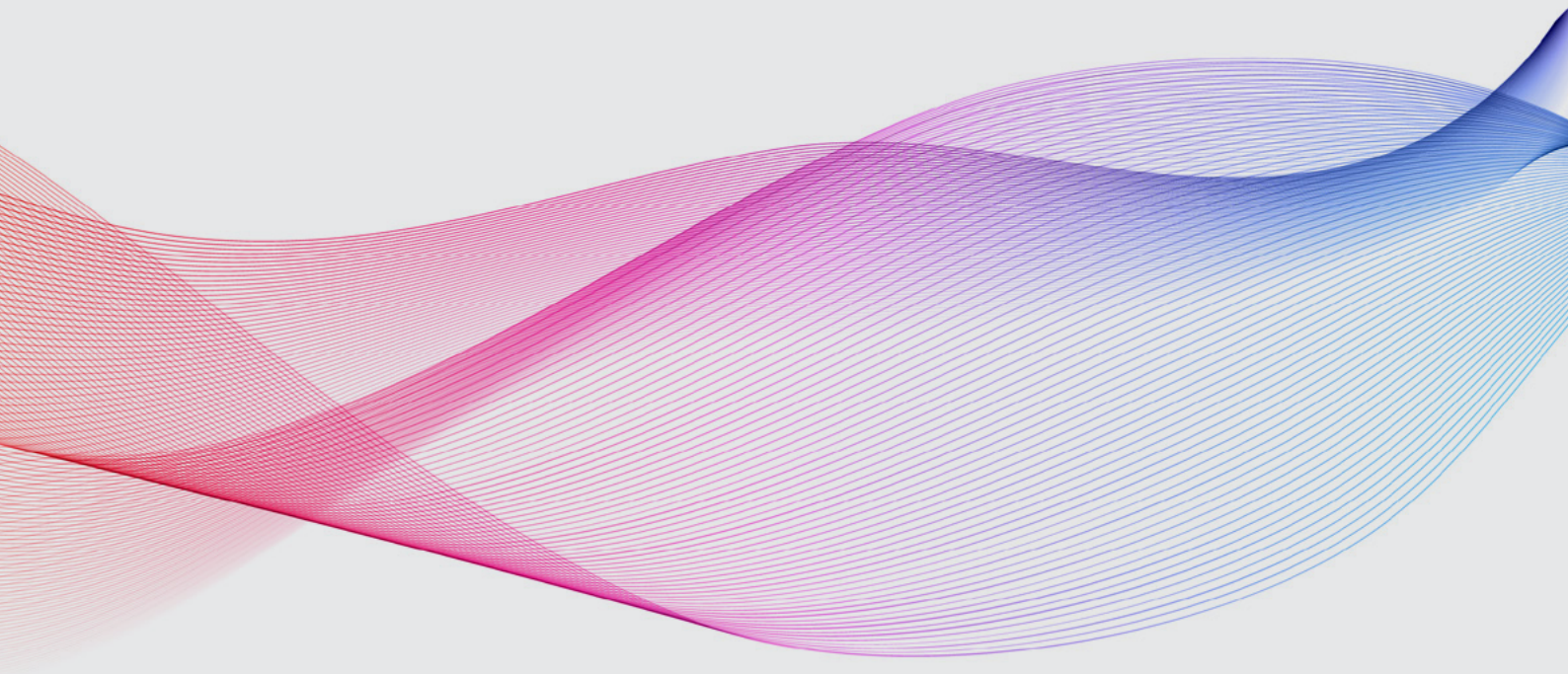
Research and service delivery have advanced through the Clinical Trials Law and large national programmes. The Gaucher initiative has trained more than 500 healthcare workers, treated over 300 patients, and provided more than 400,000 vials of therapy. The national spinal muscular atrophy programme has screened 24,000 children and offered treatment to 215 eligible patients, including 42 who received the gene injection mentioned in the transcript. For cystic fibrosis, more than 400 patients now access the treatment that includes three ingredients: elexacaftor, tezacaftor, and ivacaftor.

Despite progress, major barriers persist, including delayed diagnosis, low awareness, limited funding, geographic inequities, and high treatment costs. To address these challenges, Egypt established its first Rare Disease Fund Law in 2024, supported by a Supreme Scientific Council tasked with national registries, priority setting, and financing guidance. Furthermore, rare diseases are now included in the National Health Strategy 2024 to 2030, which focuses on equitable access, sustainable financing, stronger diagnostics, and wider treatment pathways. Current efforts emphasize awareness, national registries, expanded diagnostic capacity, and improved access to innovative therapies.

A significant milestone in 2025 was the unanimous adoption of the World Health Assembly Resolution on Rare Diseases, which mandates a ten-year Global Action Plan and reinforces global attention to this issue. Egypt's future priorities include broadening awareness, improving diagnosis and treatment access, integrating rare diseases into universal health coverage and primary care, completing national registries, strengthening the workforce, and developing sustainable reimbursement models for long-term patient access.

CHAPTER 2.

Methodology



CHAPTER 2.

Methodology

This report draws on an approach that integrates both documentary review and stakeholder perspectives. Sources of evidence include published and grey literature, health system indicators from global databases such as the WHO and the World Bank, other publicly available resources, and key informant interviews with experts and public policymakers across the region.

In addition to documentary review and in-depth key informant interviews, this report also synthesises insights emerging from the Southeast Asia Rare Diseases Policy Forum. The discussions, breakout sessions, and country presentations provided first-hand perspectives from policymakers, patient advocates, and technical experts across ASEAN.

The report is organised around four thematic areas. This framework was adapted from earlier work on rare disease management in Southeast Asia and aligned with the World Health Assembly Resolution on Rare Diseases and the proposed GAPRD.

Table 1. Report Thematic Areas and Key Components

Thematic Areas	Description
From Global Commitments to ASEAN Collaboration for Rare Diseases	• Translation of global commitments into ASEAN and national action plans • Multistakeholder collaboration to address challenges • Opportunities for joint priorities and regional solutions
Patient Journey from Diagnostics to Care	• Availability and affordability of diagnostic services. • Referral pathways and efforts to reduce delays. • Genomic capacity and data-sharing mechanisms, including registries. • Patient perspectives on diagnostic and care journeys.
Equitable Pathways to Treatment	• Availability of treatments, including orphan medicines and specialised medical foods and therapeutic nutrition products. • Harmonisation and specific fast track regulatory pathways. • Fit-for-purpose Health Technology Assessment.
Innovative and Sustainable Financing for Rare Disease	• Review of good practices in rare disease financing and their relevance for ASEAN. • Analysis of innovative models such as fiscal contribution, health impact funds, levies, charity, and multiparty contribution funds. • Assessment of feasibility for public–private partnerships and integration into social health insurance schemes.

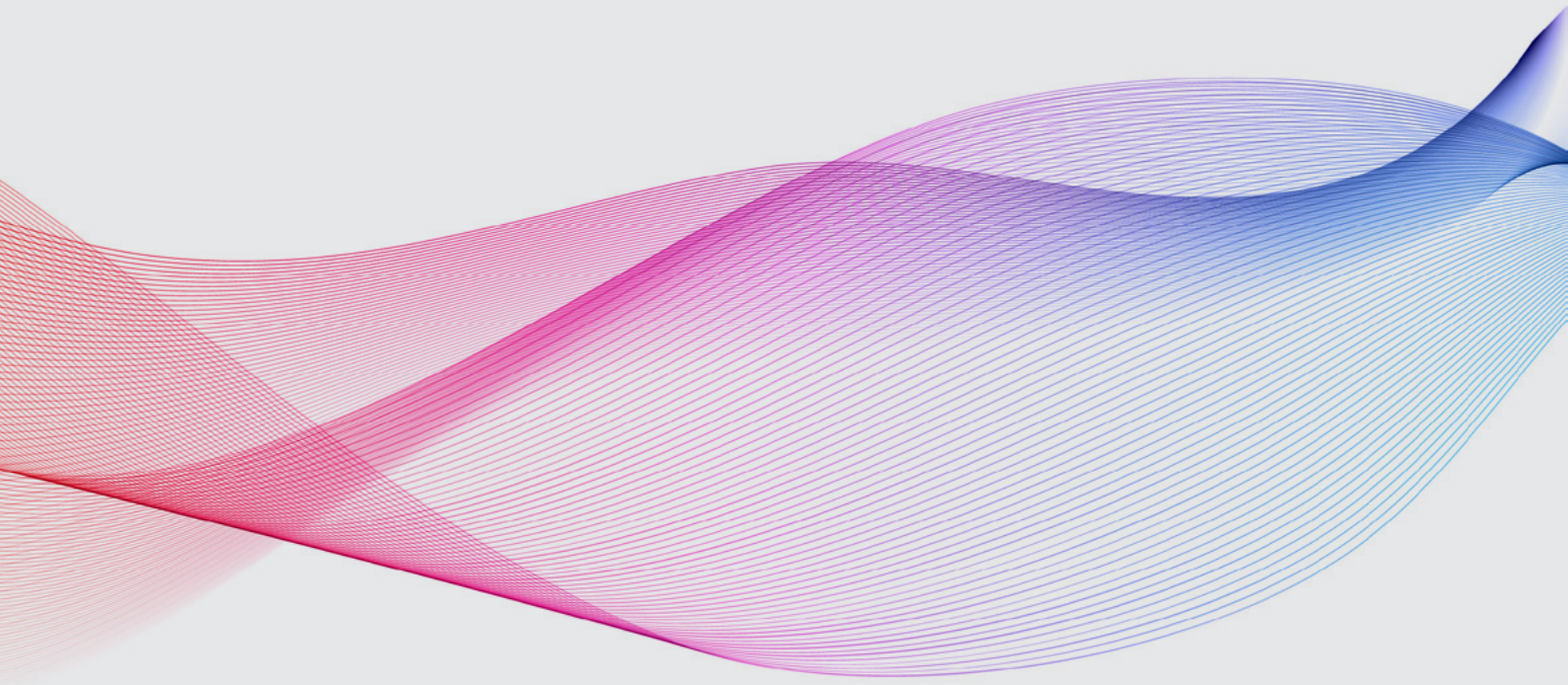
Evidence is presented thematically, using narrative synthesis and illustrative tables to highlight country experiences, regional trends, and key lessons. By triangulating literature, databases, and key informant interviews, the study ensures that findings are both evidence-based and informed by lived realities, providing a strong foundation for practical recommendations.

Limitations

The limitations of this report are acknowledged, notably the scarcity of published data, heterogeneity in definitions of rare disease, and language diversity across ASEAN countries. Nevertheless, the triangulation of literature, global databases, and key informant perspectives provides a sufficient foundation for advancing regional insights and formulating actionable recommendations.

CHAPTER 3.

Rare Disease Governance Across Several ASEAN Member States



CHAPTER 3.

Rare Disease Governance Across Several ASEAN Member States

Rare disease governance across ASEAN Member States is characterised by multi-actor institutional arrangements that span health ministries, regulatory authorities, research bodies, health insurance agencies, and specialised technical committees. While formal rare disease frameworks are at different stages of development, most countries rely on a combination of central health authorities, regulatory systems, financing institutions, and emerging genomic and research platforms to shape rare disease policy, access pathways, and service delivery. These governance structures influence how rare diseases are prioritised, how responsibilities are distributed across agencies, and how diagnostics, treatments, and long-term care are organised within national health systems.

3.1 Indonesia

In Indonesia, rare disease governance is distributed across several national institutions. The Ministry of Health serves as the central authority responsible for health policy, service delivery, referral systems, and national initiatives related to genomics and rare disease management.[21] Regulatory oversight is led by the National Agency of Drug and Food Control (BPOM), which is responsible for evaluating and approving medicines,[22] including accelerated review processes and special access mechanisms relevant to orphan drugs.

Research and innovation functions are coordinated by the National Research and Innovation Agency (BRIN), which integrates multidisciplinary research to generate evidence and support technological development, including in the field of rare diseases, to support national policy and strengthen the health system.[23] Financing and reimbursement decisions are shaped by the Social

21 Ministry of Health Indonesia. (n.d.). *Tugas dan Fungsi*. kemenkes.go.id. Link
22 BPOM. (n.d.). *Profil BPOM*. pom.go.id. Link
23 BRIN. (2025, December 26). *Profil BRIN*. Brin.go.id. Link

Security Administering Body for Health (BPJS Kesehatan), which administers the national health insurance scheme (JKN) and influences access to diagnostics, services, and medicines.[24] Technical advisory input on coverage decisions is provided by the Indonesian Health Technology Assessment Committee (InaHTAC).[25] In parallel, the Ministry of Health-led Indonesian Genomics and Biobank Initiative (BGSi) coordinates genomic laboratories, biobanking, and referral hospitals, strengthening diagnostic infrastructure increasingly relevant to rare disease detection and management.[26]

3.2 Malaysia

Malaysia has developed a more formalised rare disease governance architecture anchored within the Ministry of Health Malaysia, which oversees national health policy, service delivery, referral hospitals, laboratory and genetic services, and the overall coordination of rare disease management. This includes stewardship of the National Rare Disease Policy and leadership of the National Rare Disease Committee (NRDC), a multi-stakeholder governance body that brings together clinical, regulatory, financing, and advocacy functions.[27]

The NRDC is supported by four technical working committees covering clinical management and patient support, orphan drug management, advocacy and health education, and rare disease technology assessment. Regulatory authority for orphan medicines sits with the National Pharmaceutical Regulatory Agency (NPRA), which is responsible for designation and registration processes, supported by technical evaluation committees. Health technology assessment functions are provided by the Malaysian Health Technology Assessment Section (MaHTAS), which supports access and funding deliberations through HTA and multi-criteria decision analysis within the NRDC framework.

Beyond the health sector, the Ministry of Higher Education oversees public universities and teaching hospitals that contribute specialist services, clinical genetics expertise, and research capacity. The Social Welfare Department (Jabatan Kebajikan Masyarakat, JKM), under the Ministry of Women, Family and Community Development, complements the health system by providing social development, rehabilitation, and welfare support relevant to individuals and families affected by rare diseases.

24 Ministry of Health Indonesia. (2016, September 8). *Jaminan kesehatan nasional*. Ayo Sehat Kementerian Kesehatan RI. Link

25 Armansyah, Masytoh, L. S., Turnip, R. N., Haryani, W., Herlinawaty, E., Miftahussaadah, Dewi, R., Sari, N. M., Rosita, T., Trihartini, F., Budiman, D. R., Karlina, & Restuningtyas, F. R. (2022). *Health Technology Assessment Indonesia Annual Review 2022*. Ministry of Health Indonesia. Link

26 Ministry of Health Indonesia. (2022, August 14). *Mengenal Biomedical Genome Science Initiative (BGSI)*. kemkes.go.id. Link

27 Ministry of Health Malaysia. (2025). *National Policy for Rare Diseases in Malaysia* (MOH/P/PAK/576.25(GU)-e). Medical Development Division. Link

3.3 The Philippines

In the Philippines, rare disease governance is led by the Department of Health, which serves as the principal agency responsible for rare disease policy development, newborn screening, and service delivery. Financing support is provided through PhilHealth,[28] the national health insurance agency, which plays a central role in covering newborn screening and selected services related to rare conditions. Together, these institutions form the core governance structure for implementing the country's rare disease legislation and associated programmes.

3.4 Singapore

Singapore's rare disease governance is coordinated by the Ministry of Health, which oversees national health policy, financing, and service delivery, and provides a multi-tiered framework for supporting rare disease care. Regulatory oversight of medicines, cell therapies, and medical devices is conducted by the Health Sciences Authority (HSA).[29] Health technology assessment and clinical guidance functions are led by the Agency for Care Effectiveness (ACE), which supports evidence-informed decision making on the use of health technologies and clinical pathways.[30]

A distinctive feature of Singapore's governance model is the Rare Disease Fund (RDF), a national charitable financing mechanism that provides long-term financial support for patients requiring high-cost treatments.[31] The RDF is managed by the KK Women's and Children's Hospital Health Fund under the SingHealth Fund, with support from the Ministry of Health, forming an integrated public-philanthropic model for rare disease financing.

3.5 Thailand

Thailand's rare disease governance is anchored within the Ministry of Public Health, which provides overall stewardship of health policy and services. Financing and benefit design for rare diseases are largely administered by the National Health Security Office, which manages the Universal Coverage Scheme and associated access mechanisms.[32] Diagnostic and laboratory system strengthening is supported by the Department of Medical Sciences, which helps expand testing capacity for the detection and management of rare diseases.[33]

28 PhilHealth. (2018). *PhilHealth Circular No. 2018-00214 Enhancement of Newborn Care Package*. Philippine Star. Link
29 Health Sciences Authority. (n.d.). *About HSA*. hsa.gov.sg. Link
30 Agency for Care Effectiveness. (2025, October 27). *Who we are*. ace-hta.gov.sg. Link
31 KK Women's and Children's Hospital. (n.d.). *Rare Disease Fund*. Link
32 National Health Security Office. (2021, March 2). *24 rare diseases added to UCS But challenges remain*. Link
33 The Nation. (2025, August 31). *Medical Sciences Department opens genomic health centre for full gene testing*. Nationthailand. Link

3.6 Viet Nam

In Viet Nam, the Ministry of Health holds primary responsibility for health policy development, insurance reform, and service delivery, including areas relevant to rare diseases. Social health insurance agencies administer coverage and reimbursement mechanisms for eligible conditions and services, shaping access pathways within the national system. While formal rare disease governance structures are still evolving, these institutions form the core architecture through which rare disease policy and access issues are addressed.[34]

Table 2. Key Governance Stakeholders for Rare Diseases

Indonesia	Malaysia	The Philippines
• Ministry of Health • BPOM • BRIN • BPJS Kesehatan • InaHTAC • BGSi	• Ministry of Health • Ministry of Higher Education • Welfare Department • NRDC • NPRA • MaHTAS	• Department of Health • PhilHealth
Singapore	**Thailand**	**Viet Nam**
• Ministry of Health • Rare Disease Fund • HSA • ACE	• Ministry of Public Health • NHSO • Department of Medical Sciences	• Ministry of Health • Social Health Insurance Agencies

34 Ministry of Finance Viet Nam. (2023, July 21). *Health Insurance - The "golden" card for public health*. Link

CHAPTER 4.

ASEAN Leadership in Advancing Regional Collaboration on Rare Diseases

CHAPTER 4.

ASEAN Leadership in Advancing Regional Collaboration on Rare Diseases

4.1 Global Commitments on Rare Diseases

A major turning point occurred in 2019 when joint advocacy efforts secured the inclusion of rare diseases in the **United Nations Political Declaration on Universal Health Coverage**, framing them as issues of equity and inclusion.[35,36] These efforts culminated in significant global milestones: the **2021 United Nations General Assembly Resolution on "Persons Living with a Rare Disease and Their Families"** and the landmark **2025 World Health Assembly Resolution**. The UN General Assembly Resolution 76/132 in 2021 marked the first political acknowledgement that rare diseases are medical, social and economic issues that require systemic inclusion in the 2030 Agenda. Momentum increased in February 2025 when the **WHO Executive Board** recommended adoption of the Resolution on Rare Diseases, which represented a people-centred, equity-driven approach to rare diseases. In May 2025, this was followed by the unanimous adoption of the **WHA Resolution 78.11**, which called for integrating rare diseases into UHC, strengthening diagnostics and registries, improving equitable access to medicines, and mandating the development of a **10-year Global Action Plan.**

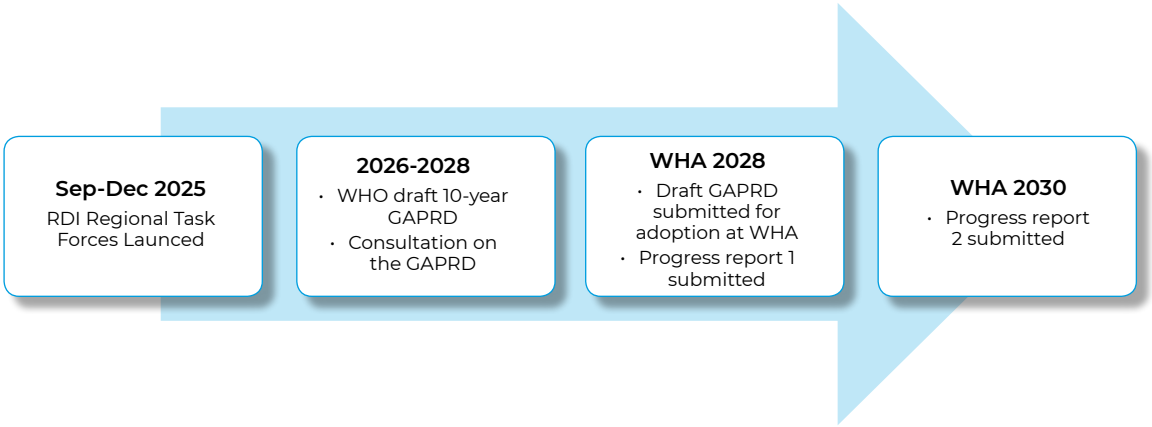
These WHO global frameworks frame rare diseases not as isolated clinical conditions, but as a **health-systems and equity challenge** that intersects with UHC, people-centred service delivery, chronic disease management, and the responsible use of genomics. They reinforce the need for integrated care pathways, sustained and equitable financing, strong primary and referral

35 International Rare Diseases Research Consortium. (n.d.). *Vision and goals*. IRDiRC. Link
36 World Health Organization. (n.d.). UN high-level meetings on universal health coverage WHO. Link

systems, and robust governance of data and technology. For ASEAN Member States, alignment with these global commitments provides a coherent policy foundation to reduce diagnostic delays, improve continuity of care, protect households from financial hardship, and ensure that advances in genomics translate into equitable public health benefit rather than widened disparities.

The adoption of WHA Resolution 78.11 has also initiated a structured process to guide its implementation at global, regional, and national levels. Beginning in late 2025, **RDI's Regional Task Forces** began to established to support early coordination and facilitate stakeholder engagement. WHO is expected to lead the development of a ten-year **GAPRD between 2026 and 2028** through a formal consultation process involving member states and relevant stakeholders. The draft plan is anticipated for consideration at the **2028 World Health Assembly,** accompanied by an initial progress report, with a subsequent **progress report scheduled for 2030**. This timeline provides a clear framework for sustained follow-through on the resolution and for supporting countries as they integrate rare diseases into broader health-system strengthening efforts.[37]

Picture 2. Upcoming Milestones in the Global Rare Disease Agenda[38]



Together, these commitments are anchored in a broader set of WHO policy frameworks that articulate how equity, people-centred care, chronic disease management, and genomics should be operationalised within health systems, as summarised below.

These WHO global frameworks frame rare diseases as a health-systems and equity challenge linked to universal health coverage, people-centred care, chronic disease management, and responsible use of genomics. Across ASEAN Member States, progress is emerging across varied economic and health system contexts through policy and legal frameworks, integration into UHC

37 Rare Disease International. (n.d.). *The WHA Resolution on Rare Diseases*. RDI. Link
38 Ibid.

benefits, expansion of genomic and screening capacity, and strengthened referral pathways. Building on these experiences**, ASEAN Member States are well placed to contribute global south-informed implementation insights to the development and operationalisation of the WHO ten-year GAPRD**, particularly to reduce diagnostic delays, improve continuity of care, and strengthen financial protection.

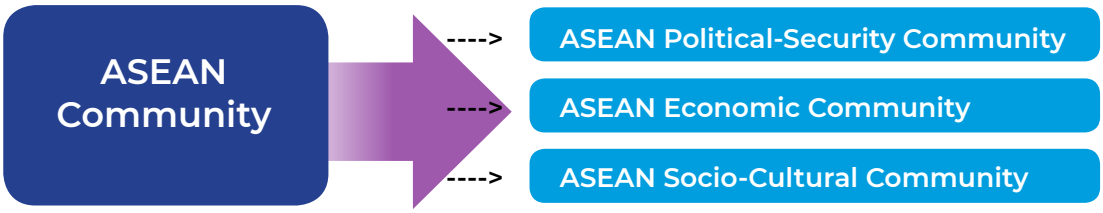
DID YOU KNOW?

The global rare disease policy landscape has developed relatively recently. Key milestones include the establishment of Orphanet by INSERM and EURORDIS in 1997, followed by the adoption of the European Union's Orphan Medicinal Products Regulation in 1999–2000 and the launch of France's first National Plan for Rare Diseases in 2004. International visibility and policy coordination expanded with the inauguration of Rare Disease Day by EURORDIS in 2008 and the issuance of the EU Council Recommendation on rare diseases in 2009. Subsequent advances included the creation of the European Reference Networks and the International Rare Diseases Research Consortium (IRDiRC) in 2011, the establishment of the UN NGO Committee for Rare Diseases in 2014, and the formation of Rare Diseases International in 2015. By 2017, 24 European Reference Networks had been formally launched, representing a major step in structured cross-border collaboration for rare disease care and research.

4.2 ASEAN-Led Alignment of Regional Priorities with Global Commitments on Rare Diseases

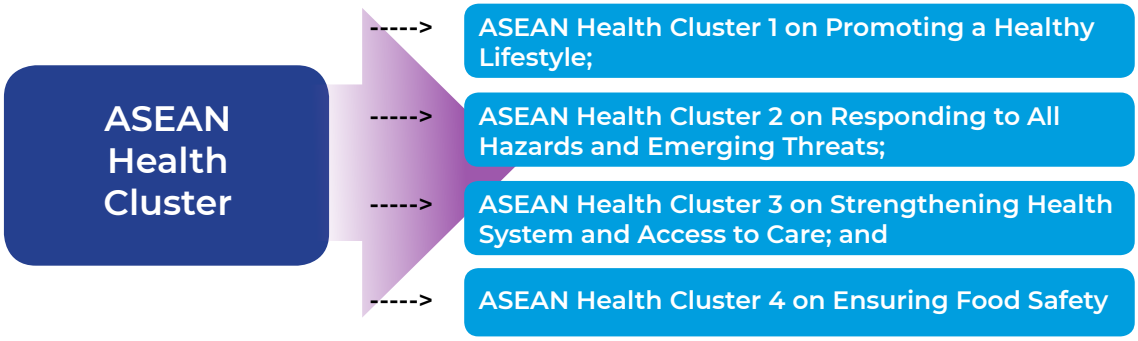
ASEAN's community agenda was formally articulated in 2003 through the Declaration of ASEAN Concord II (Bali Concord II), which set out the vision of an ASEAN Community founded on three closely interconnected and mutually reinforcing pillars: political and security cooperation, economic integration, and socio-cultural cooperation, with the aim of securing lasting peace, stability, and shared prosperity in the region.[39] This agenda was subsequently reinforced by the adoption of the ASEAN Charter signed in 2007, which provided a legal and institutional basis for ASEAN's evolution into a rules-based organisation.[40] A major milestone was reached in 2015 with the adoption of the Kuala Lumpur Declaration on "ASEAN 2025: Forging Ahead Together" and the three ASEAN Community Blueprints, which formally established the ASEAN Community on 31 December 2015.[41]

39 ASEAN Secretariat. (2012, May 11). *Declaration of ASEAN Concord II (Bali Concord II).* Link
40 Association of Southeast Asian Nations. (2008). *The ASEAN Charter*. ASEAN Secretariat. Link
41 ASEAN Secretariat. (2015). *ASEAN 2025: Forging Ahead Together.* Association of Southeast Asian Nations. Link



The ASCC is the pillar of the ASEAN Community responsible for advancing human development, social well-being, and quality of life across the region. Health is a central area of cooperation under the ASCC, alongside education, social welfare, labour, and environmental sustainability. Through the ASCC and its Blueprint 2025,[42] ASEAN promotes regional collaboration to strengthen health systems, expand universal health coverage, improve access to essential health services, and build collective capacity to prevent and respond to health-related risks, including outbreaks, disasters, and other cross-border health threats. The ASCC also supports cooperation on maternal and child health, nutrition, ageing, disability, and the protection of vulnerable populations, recognising the close links between health outcomes and broader social conditions. This socio-cultural framework provides ASEAN with an established regional platform to address complex and long-term health challenges that require sustained cooperation, shared learning, and coordinated policy approaches, including the growing burden of non-communicable and rare diseases.

The ASEAN Health Sector anchors health cooperation under the ASCC. It is organised around four priority areas, known as the ASEAN Health Clusters, as defined in the ASEAN Post-2015 Health Development Agenda:[43]



ASEAN Post-2015 Health Development Agenda (2021 to 2025), particularly Cluster 3 on strengthening health systems and access to care, provides a natural basis for future rare-disease cooperation through its focus on equity, innovation, and people-centred health services. Cluster 1 on Promoting Healthy Lifestyles also offers relevant entry points, as its emphasis on health promotion, community

42 ASEAN Secretariat. (2016). *ASEAN Socio-Cultural Community Blueprint 2025*. Association of Southeast Asian Nations. Link

43 ASEAN Secretariat. (2023). *ASEAN Post-2015 Health Development Agenda (APHDA) 2021-2025*. ASEAN. Link

engagement, and early detection aligns with efforts to raise awareness of rare diseases and improve timely care-seeking. Taken together, these clusters illustrate a clear progression from global recognition to operational mandate, positioning ASEAN to translate global commitments into coherent regional and national action.[44]

Picture 3. Primary Health Care as a Catalyst for Rare Disease Management

Furthermore, rare disease care can be embedded into primary health centres by strengthening their role in early recognition, referral, and long-term follow-up. Frontline providers should be supported to identify unusual or persistent symptoms and to use clear criteria for referring patients to specialised diagnostic centres, rather than trying to make a diagnosis themselves. Simple, well-defined referral pathways between primary care and tertiary hospitals are essential to shorten time to diagnosis. After a diagnosis is confirmed, primary health centres can help coordinate ongoing care by supporting routine follow-up, treatment adherence, rehabilitation, and family support in collaboration with specialists. Where formally regulated, Traditional and Complementary Medicine can be used at primary care level as a supportive service to improve symptom relief and overall well-being, while remaining aligned with medical care plans.[45]

Table 3. ASEAN Potential in Advancing Global Commitments on Rare Diseases

ASEAN Leaders' Declaration on the One Health Initiative	Provides a high-level political framework to strengthen cross-sectoral and cross-border collaboration, which is essential for improving rare disease management across ASEAN.
ASEAN Declaration of Commitment on Drug Security and Self-Reliance (ADSSR)	Reinforces regional cooperation on access, manufacturing, and supply resilience, creating an enabling foundation to improve the availability of orphan drugs and specialised therapies in ASEAN.
ASEAN Post-2015 Health Development Agenda 2021–2025 (APHDA)	Offers a strategic regional health roadmap under which rare disease priorities can be integrated into broader commitments on UHC, health systems strengthening, and equitable access.

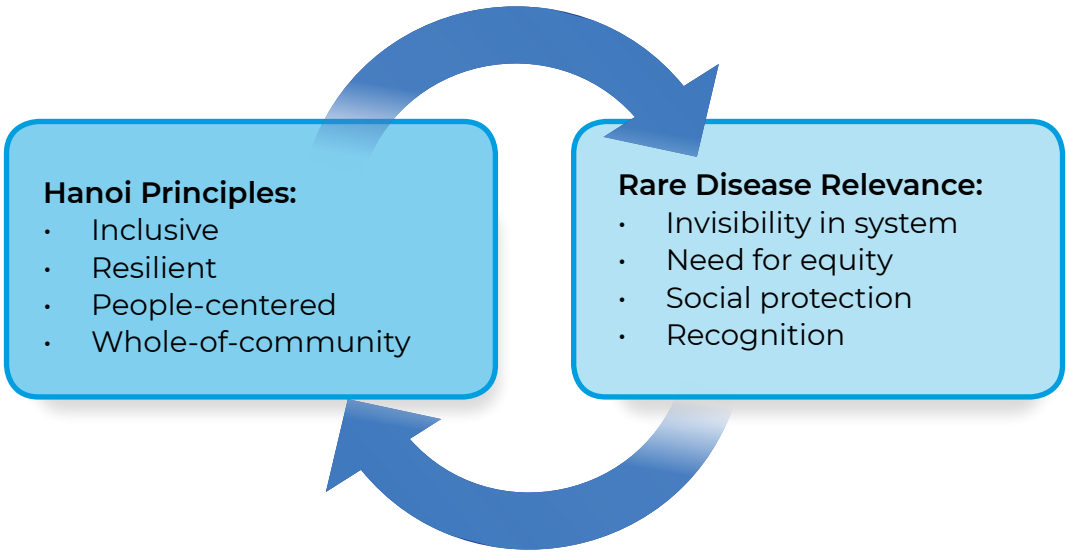
44 United Nations. (2022). Addressing the challenges of persons living with a rare disease and their families. In *United Nations* (Report A/RES/76/132; pp. 2–5). Link

45 ASEAN Secretariat. (2023). *Joint Report on the Situational Analyses of ASEAN Primary Health Care and Traditional and Complementary Medicine Capacities: Towards the Enhancement of Quality Health Care through Primary Health Care Capacities in ASEAN Member States*. Link

ASEAN Pharmaceutical Product Working Group (PPWG)	Serves as a key technical platform to advance regulatory convergence and reliance mechanisms that can accelerate the registration and regional availability of rare disease medicines.
Joint Assessment Coordinating Group (JACG)	Supports collaborative regulatory and assessment processes, enabling more efficient joint or coordinated reviews that are particularly relevant for expediting access to orphan products in resource-constrained settings.

The Hanoi Declaration on the ASEAN Community's Post-2025 Vision (2020) sets the region's direction for an inclusive, resilient, and people-centred ASEAN grounded in equity, sustainability, and whole-of-society engagement. Although rare diseases are not explicitly mentioned, its focus on inclusion, social resilience, and leaving no one behind aligns closely with the global rare-disease agenda. As ASEAN develops its post-2025 vision, incorporating rare diseases would strengthen the region's commitment to ensuring that even small and often invisible patient groups benefit from regional cooperation. Furthermore, the ASEAN Pharmaceutical Product Working Group (PPWG) and ASEAN Joint Assessment Coordinating Group (JACG) offer existing regulatory platforms that can support this ambition by providing harmonised standards and joint assessment pathways for rare disease treatments across ASEAN (*see annexure*).[46]

Picture 4. Alignment Between the Hanoi Principles and Rare Disease Priorities



46 ASEAN Secretariat. (2020). Ha Noi Declaration on the ASEAN Community's Post-2025 Vision. In *ASEAN*. Link

The ASEAN region presents a diverse policy landscape for rare diseases, reflecting varying levels of institutional commitment, health system maturity, and stakeholder participation. Across the six focus countries, there is gradual progress toward integrating rare diseases into national health agendas, though challenges persist in financing, registries, and equitable service delivery (*see annexure*).

Table 4. Challenges and Opportunities

Challenges	Opportunities
 **Limited Integration of Rare Diseases into ASEAN's Health Agenda**	Opportunities include embedding rare diseases within existing health clusters, creating a regional coordination mechanism, using established ASEAN platforms for alignment and knowledge exchange, promoting joint research and capacity-building, updating national frameworks with shared indicators, and advancing harmonised registries, data standards, and referral arrangements.
 **Delayed Early Detection and Insufficient Diagnostic Capacity**	Progress can be supported through broader newborn screening, the gradual integration of genomic testing, development of national registries, strengthened laboratory and bioinformatics capacity, engagement with public–private innovations, and exploration of emerging approaches for presymptomatic and carrier screening.
 **Unequal Pathways to Treatment**	Opportunities lie in wider use of telemedicine and AI, the development of centres of excellence, greater incorporation of patient-centred policies and participation, inclusion of quality-of-life and social inclusion measures, and awareness efforts that help reduce stigma and encourage timely care-seeking.
 **High Financial Burden and Unsustainable Funding**	Potential pathways include the gradual inclusion of rare diseases within UHC packages, the use of transparent prioritisation processes, exploration of innovative financing options and blended funds, and consideration of pooled procurement to broaden access to high-cost therapies.
 **Evolving National Governance and Limited Multi-sectoral Coordination**	National governance may be strengthened through dedicated units within health ministries, closer links with education, social protection and research sectors, and structured involvement of patients, families, and caregivers in policy and programme development.

Together, these five areas represent actionable starting points for collective action. They build on ASEAN's long-standing principles of voluntary cooperation, mutual learning, and shared responsibility. Early progress on data consistency, diagnostic quality, access facilitation, patient participation, and joint monitoring would demonstrate that meaningful regional collaboration is both possible and beneficial, even with modest resources. Step by step, these efforts can lay the groundwork for more comprehensive regional frameworks that enhance equity and care for people living with rare diseases across ASEAN.

ASEAN Opportunities for Synergy

Across ASEAN, rare disease policy and system development has evolved along two overlapping but unequal strategic streams.

- **Stream A: Population-anchored early detection, diagnosis, and prevention**, focus on foundational platforms such as newborn screening, confirmatory diagnostics, genetic counselling, and targeted prevention in high-risk subgroups, where cost-effectiveness, equity, and population-level health impact are demonstrable. This stream reflects a systems-oriented approach that strengthens core diagnostic and referral capacity while benefiting broader population health.
- **Stream B: Selective access to high-cost innovative therapies** refers to structured pathways for a limited number of feasible treatments, including haemophilia, selected lysosomal storage disorders, and spinal muscular atrophy, where compassionate access, partial public financing, negotiated pricing, or special access schemes can be mobilised within acknowledged affordability constraints. While narrower in reach, this stream responds to urgent unmet clinical need and has often served as an entry point for rare disease policy development in the region. Together, these overlapping streams provide the strategic lens through which the regional priority areas outlined in this chapter are framed.

In response to shared constraints in capacity, financing, and collective capacity, opportunities for ASEAN collaboration can be understood across three progressive and complementary tiers, reflecting varying levels of feasibility, political sensitivity, and system readiness among Member States.

- **Tier 1 — Sharing** focuses on the exchange of knowledge and experience, including policy models, diagnostic and referral algorithms, health technology assessment approaches, and governance frameworks. This tier represents the most immediately actionable form of collaboration and can

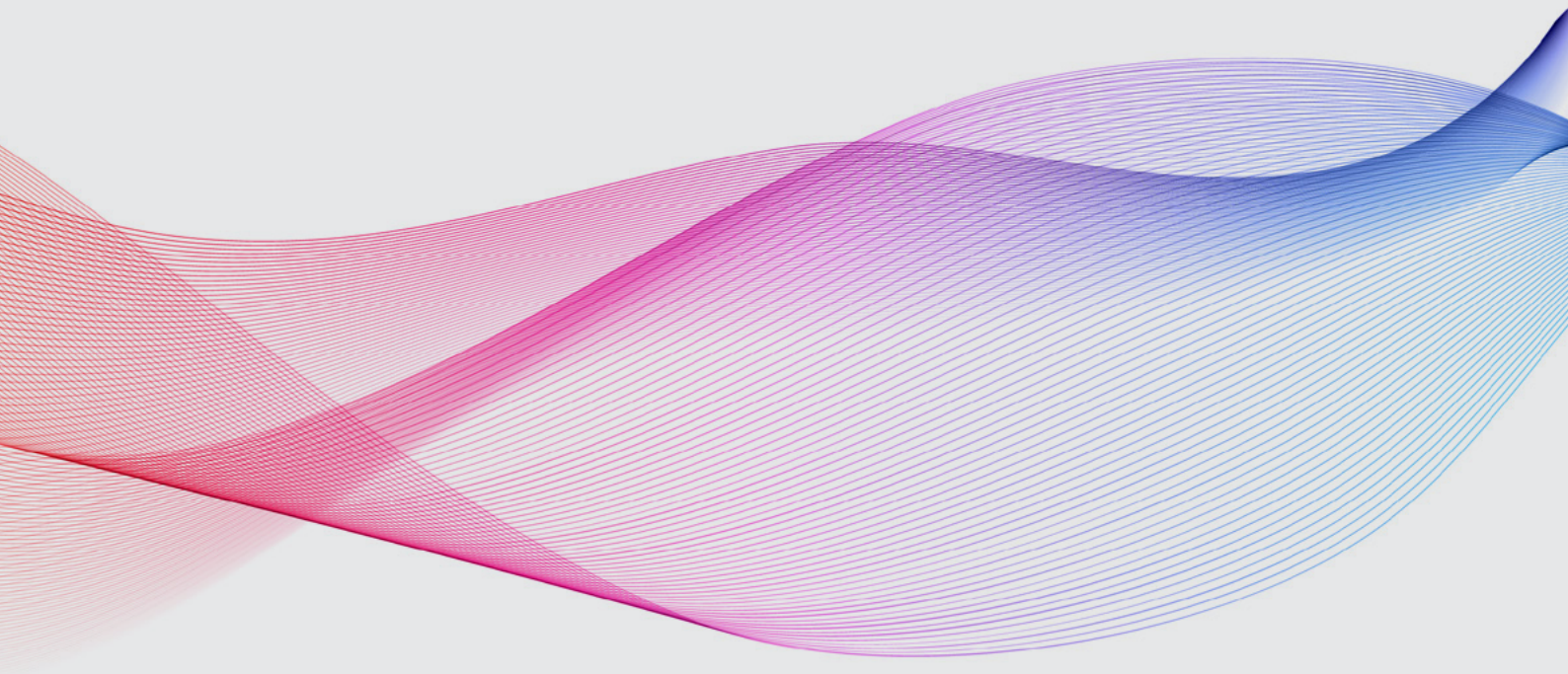
support learning, policy convergence, and capacity strengthening without requiring formal harmonisation.

- **Tier 2 — Cooperation** builds on shared learning through more structured forms of engagement, such as joint workforce training initiatives, harmonisation of rare disease datasets and coding systems (including ORPHA and ICD), bilateral or subregional diagnostic referral arrangements, and the development of quality assurance networks. These cooperative actions can enhance efficiency and consistency while remaining adaptable to national contexts.
- **Tier 3 — Collaboration** reflects longer-term and aspirational mechanisms that may be pursued selectively as trust, capacity, and political alignment deepen. These include pooled procurement pilots for selected therapies, shared genomic or reference laboratory hubs, and regional platforms for dialogue and negotiation, particularly where collective approaches may improve affordability and access.

ASEAN rare disease systems remain fragmented and uneven across Member States, yet there are clear signs of progress in several countries, including the expansion of newborn screening, the development of specialised services, regulatory improvements, and emerging financing mechanisms. Embedding rare disease priorities within existing ASEAN health cooperation mechanisms and advancing an ASEAN Rare Disease Declaration and Action Plan would institutionalise regional collaboration, strengthen coordination, and support more systematic national reforms. The two-stream framing and an ASEAN-specific roadmap provide a practical foundation for action that reflects regional contexts, supports gradual health system strengthening, and aligns with broader global health commitments.

CHAPTER 5.

Patient Journey from Diagnostics to Care



CHAPTER 5.

Patient Journey from Diagnostics to Care

5.1 Availability and Affordability of Diagnostic Services

Across the six ASEAN countries reviewed, the availability and affordability of diagnostic services for rare conditions are advancing but unevenly. The diagnostic experience for people living with rare diseases is shaped by multiple entry points, fragmented pathways, and uneven system capacity, rather than a single, linear patient journey. Families typically enter the health system through one of two routes: population-based newborn screening, where it exists, or symptom-led clinical presentation, which remains the dominant pathway for most rare conditions across the region.

NBS functions as a population-level triage mechanism for a limited set of conditions, enabling early identification only where coverage, panel scope, and follow-up systems are sufficiently developed. While NBS represents an important entry point in some ASEAN countries, its reach and integration into care vary substantially. In contrast, symptom-led clinical diagnosis remains the primary pathway for most rare diseases across ASEAN, particularly in countries with limited NBS coverage or narrow screening panels. Children and adults often present with developmental delay, seizures, metabolic crises, neuromuscular symptoms, or multi-system disease, entering care through general paediatrics or specialty services.[47] In these cases, diagnosis depends heavily on clinician awareness, access to appropriate testing, and the ability to navigate referral systems.

Singapore and Thailand illustrate relatively mature models, combining near-universal screening with subsidised confirmatory testing and access to advanced laboratories. The Philippines shows substantial gains through an expanded panel supported by PhilHealth, though confirmatory testing and follow-up are not fully financed. Malaysia's policy commitments signal expansion, but current capacity constraints and the cost of specialised tests still push some families toward private or overseas options. Viet Nam's services are

47 Yang, D., Ren, X., Lu, Y., & Han, J. (2021). Current diagnosis and management of rare pediatric diseases in China. *Intractable & rare diseases research, 10*(4), 223–237. Link

growing, but coverage and reimbursement remain limited and concentrated in major cities. Indonesia is in transition, with national congenital hypothyroidism screening not yet reaching high coverage and broader panels still piloted locally. Across settings, services and specialists cluster in urban centres. At the same time, financing arrangements frequently cover screening but not the whole cascade of confirmatory testing, longitudinal monitoring, and genetic workups, creating equity gaps for low-income and remote households.

Indonesia

Indonesia's NBS programme remains in transition. While Minister of Health Regulation No. 78/2014 mandated screening for congenital hypothyroidism (CH), the programme's expansion beyond CH has remained mainly in pilot and research phases rather than full national implementation.[48] ACongenital hypothyroidism had a screening coverage of 4.37% in 2023, and efforts to expand screening are ongoing and noted that in 2023, the Ministry of Health tied CH screening to insurance claim eligibility for newborn delivery payments.[49] Pilot studies have tested CAH screening (using 17-OHP methods) in several cities and recorded positive recall and confirmation rates, but these remain local trials rather than mandated national services.[50] Implementation challenges are well documented: newborns discharged too early, limited training, supply issues (e.g., filter paper quality), and low follow-up adherence. As of mid-2024, geographic variation in uptake remains substantial, indicating that nationwide, consistent expansion has not yet been achieved. Future policy documents or official MOH releases are needed to confirm full integration into JKN or official adoption of expanded panels across all provinces.

Malaysia

Malaysia's newborn screening programme is implemented through public hospitals and currently includes:[51]

1. Congenital hypothyroidism, and
2. G6PD deficiency.

In addition, the Institute for Medical Research (IMR) has led a pilot initiative using tandem mass spectrometry to screen for inborn errors of metabolism

48 Octavius, G. S., Daleni, V. A., & Sagala, Y. D. S. (2023). An Insight into Indonesia's Challenges in Implementing Newborn Screening Programmes and Their Future Implications. *Children*, *10*(7), 1216. Link

49 Ministry of Health Indonesia. (2023, July 1). Skrining Bayi Baru Lahir di Indonesia [Slide show]. IACC. Link

50 Pulungan, A. B., Soesanti, F., Utari, A., Pritayati, N., Julia, M., Annisa, D., Andarie, A. A., & Bikin, I. W. (2020). Preliminary study of newborn screening for congenital hypothyroidism and congenital adrenal hyperplasia in Indonesia. *DOAJ (DOAJ: Directory of Open Access Journals)*. Link

51 Ministry of Health Malaysia. (2025). *National Policy for Rare Diseases in Malaysia* (MOH/P/PAK/576.25(GU)-e). Medical Development Division. Link

(IEM)[52]. The recently launched NPRD 2025 includes commitments to expand early diagnostic capacity, establish national genetic reference laboratories, and integrate screening results into electronic medical records. Although diagnostic tests in public facilities are heavily subsidised, limited laboratory capacity and a shortage of clinical geneticists lead to referrals abroad or to private centres, which can be costly. Families often rely on patient-organisation fundraising to cover sequencing or enzyme assays. The Institute for Medical Research, however, provides comprehensive nationwide confirmatory testing for inborn errors of metabolism, contributing an important national capability despite ongoing needs to strengthen diagnostic resources more broadly.

The Philippines

The Philippines has made significant progress in expanding access to diagnostic services for rare diseases through its expanded newborn blood-spot screening (ENBS) programme, now covering 29 conditions, including metabolic, endocrine, and hematologic disorders.[53] The ENBS programme enables early identification of several serious and potentially life-threatening rare conditions, including:[54]

Table 5. Expanded Newborn Blood-Spot Screening Conditions Covered in the Philippines

1. Primary Congenital Hypothyroidism
2. Congenital Adrenal Hyperplasia (21-Hydroxylase Deficiency)
3. Homocystinuria
4. Methionine Adenosine Transferase Deficiency (Hypermethioninemia
5. Maple Syrup Urine Disease
6. Phenylketonuria
7. Tyrosinemia Type I, II, III
8. Carnitine Palmitoyltransferase I Deficiency
9. Carnitine Palmitoyltransferase II Deficiency
10. Carnitine Uptake Deficiency
11. Glutaric Acidemia Type II
12. Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency
13. Medium Chain-Acyl-CoA Dehydrogenase Deficiency
14. Very Long Chain-Acyl-CoA Dehydrogenase Deficiency
15. Tri-functional Protein Deficiency
16. 3-Methylcrotonyl CoA Carboxylase Deficiency
17. Beta Ketothiolase Deficiency
18. Glutaric Acidemia Type I

52 Yunus ZM, Rahman SA, Choy YS, Keng WT, Ngu LH. *Pilot study of newborn screening of inborn error of metabolism using tandem mass spectrometry in Malaysia: outcome and challenges.* Journal of Pediatric Endocrinology and Metabolism. 2016;29(9):1031–1039. doi:10.1515/jpem-2016-0028. PMID: 27544719. Link

53 Padilla, C. D., Therrell, B. L., Alcausin, M. M. L. B., Chiong, M. a. D., Abacan, M. a. R., Reyes, M. E. L., Jomento, C. M., Dizon-Escoreal, M. T. T., Canlas, M. a. E., Abadingo, M. E., Posecion, J. E. W. C., Abarquez, C. G., Andal, A. P., Elizaga, A. L. G., Halili-Mendoza, B. C., Otayza, M. P. V. K., & Millington, D. S. (2022b). Successful implementation of expanded newborn screening in the Philippines using tandem mass spectrometry. *International Journal of Neonatal Screening*, *8*(1), 8. Link

54 ParenTeam. (2025, May 30). *Newborn Screening 101: What First-Time Parents Need to Know.* Link

19. Isovaleric Acidemia
20. Methylmalonic Acidemia
21. Multiple Carboxylase Deficiency
22. Propionic Acidemia
23. Citrullinemia
24. Argininosuccinic Aciduria
25. All Detectable Hemoglobinopathies and Thalassemias
26. Galactosemia
27. Glucose-6-Phosphate Dehydrogenase Deficiency
28. Cystic Fibrosis
29. Biotinidase Deficiency

The DOH operates ENBS in partnership with the National Institute of Health, supported by the PhilHealth Newborn Screening Benefit Package. Coverage reached roughly 92% of live births by 2024, a sharp increase from 60% a decade earlier.[55] Diagnostic affordability is bolstered by PhilHealth reimbursement, which eliminates out-of-pocket costs for the initial screening. Diagnostic affordability is therefore significantly strengthened at the screening stage, as families do not pay out of pocket for the initial test. However, confirmatory testing and follow-up laboratory procedures are often not fully covered, which continues to place financial pressure on low-income households. Specialised confirmatory laboratories remain concentrated mainly in Metro Manila, limiting equitable access, although the national network of Newborn Screening Continuity Clinics and regional testing centres continues to expand to improve follow-up care and referral.

Singapore

Singapore operates one of the most comprehensive rare disease diagnostic ecosystems in Southeast Asia. Universal newborn screening, introduced nationally in 2006 and progressively expanded, now covers more than 40 disorders, including:[56]

Table 6. Universal Newborn Screening Disorder Coverage in Singapore

1. Phenylketonuria including biopterin defects
2. Maple Syrup Urine Disease
3. Citrullinaemia type 1
4. Argininosuccinic Aciduria
5. Tyrosinaemia type 1
6. Homocystinuria (pyridoxine unresponsive)
7. Propionic acidaemia
8. Methylmalonic acidaemia (MUT)
9. Cobalamin A/B
10. Isovaleric acidaemia
11. ß-ketothiolase deficiency
12. Glutaric acidaemia type 1
13. Malonic aciduria
14. 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency

55 Mendoza, V. G. (n.d.). *Expanded newborn screening in the Philippines: A 2024*. Newborn Screening. Link
56 Tan, J. (2025, July 26). Singapore's national screening programme tests newborns for metabolic and heritable diseases. *The Straits Times*. Link

15. Multiple carboxylase deficiency
16. Primary carnitine deficiency / Carnitine uptake deficiency
17. Medium chain acyl-CoA dehydrogenase deficiency
18. Very long chain acyl-CoA dehydrogenase deficiency
19. Long chain hydroxy acyl-CoA dehydrogenase
20. Trifunctional protein deficiency
21. Hyperphenylalanine
22. Argininase deficiency
23. Citrin deficiency
24. Hypermethioninaemia
25. Tyrosinaemia Types 2 and 3
26. 3-Methylcrotonyl-CoA carboxylase deficiency
27. 2-Methyl-3-hydroxy butyric aciduria
28. 3-Methylglutaconyl-CoA dehydratase deficiency
29. Isobutyryl-CoA dehydrogenase deficiency
30. 2-Methylbutyryl-CoA dehydrogenase deficiency / short branch chain acyl-CoA
31. Ethylmalonic encephalopathy
32. Cobalamin C/D
33. Carnitine palmitoyltransferase deficiency type 1
34. Carnitine palmitoyltransferase deficiency type 2
35. Carnitine-acylcarnitine translocase deficiency
36. Multiple acyl-CoA dehydrogenase deficiency / glutaric aciduria type 2
37. Short chain acyl-CoA dehydrogenase deficiency
38. Medium/Short chain hydroxy acyl-CoA dehydrogenase deficiency
39. Medium chain ketoacyl-CoA thiolase deficiency
40. Galactosemia
41. Cystic Fibrosis
42. Biotinidase Deficiency
43. Congenital Adrenal Hyperplasia
44. Severe Combined Immune Deficiency

The programme is coordinated by KK Women's and Children's Hospital (KKH) in partnership with the Ministry of Health and currently screens around 90% of annual live births, equivalent to approximately 40,000 newborns each year. Expanded panels now include congenital adrenal hyperplasia, galactosaemia, biotinidase deficiency, cystic fibrosis, severe combined immunodeficiency, and a broad range of inborn errors of metabolism using tandem mass spectrometry (MS/MS). Although these conditions are individually rare, some categories of metabolic disorders together may occur at a rate of 1 in 3,000 to 3,500 births, underscoring the public health importance of population-level screening.[57]

Diagnostic affordability is supported by Singapore's tiered subsidy system, which enables citizens to access heavily subsidised confirmatory and follow-up testing at public hospitals. KKH established a dedicated Biochemical Genetics Laboratory alongside the newborn screening programme to provide confirmatory testing, diagnostic services for inborn errors of metabolism, and long-term monitoring for affected patients. Additional financial and social support is provided through the Rare Disease Fund and hospital-based

57 KK Women's and Children's Hospital. (n.d.). *Biochemical Genetics and NENS Laboratory*. kkh.com.sg. Link

charity programmes, which assist eligible families with the costs of diagnosis and treatment. Together, this integrated screening, laboratory, and financing infrastructure enables early detection, timely referral, and improved clinical outcomes for infants identified with rare and inherited disorders.[58]

Thailand

Thailand's Universal Coverage Scheme (UCS) has incorporated newborn screening and confirmatory testing into the national benefit package. The National Health Security Office (NHSO) and the Department of Medical Sciences expanded nationwide screening using tandem mass spectrometry (MS/MS), which enables the detection of more than 40 inborn errors of metabolism and other serious congenital conditions, including 24 rare diseases prioritised under the UCS benefit package.[59] According to national implementation data, newborn screening coverage has now reached approximately 98% of live births, including in non-urban regions, reflecting near-universal programme reach.[60,61,62] Screening is provided free of charge at UCS facilities, and systems have been established for rapid recall and referral within 24-48 hours for infants with positive screening results.

However, confirmatory testing and treatment are reimbursed only for conditions and medicines included in the National List of Essential Medicines.[63] This restriction limits financial protection for patients with rare diseases not explicitly listed in the NLEM, who may therefore face out-of-pocket costs. While the NHSO provides transportation and referral support to designated hospitals, gaps persist in financial coverage and the geographic distribution of specialist services, particularly outside Bangkok.[64]

58 National Centre for Infectious Diseases. (2025, October 2). *Schemes and subsidies*. NHG Health. Link

59 National Health Security Office. (2023, September 5). *Tandem mass spectrometry newborn screening is available nationwide*. NHSO. Link

60 Wichajarn, K., Sawatjui, N., Prasongdee, P., Panklin, A., Sornkayasit, K., Chungkanchana, N., Tessiri, S., Wintachai, P., Dechyotin, S., Pasomboon, C., Ratanapontee, J., Thanakitsuwan, S., & Rattanathongkom, A. (2025). The Establishment of Expanded Newborn Screening in Rural Areas of a Developing Country: A Model from Health Regions 7 and 8 in Thailand. *International Journal of Neonatal Screening*, *11*(2), 26. Link

61 Therrell, B. L., Padilla, C. D., Abadingo, M. E., et al. (2025). *Consolidated newborn bloodspot screening efforts in developing countries in the Asia Pacific—2024. International Journal of Neonatal Screening, 11*(1), Article 2. Link

62 Therrell, B. L., Padilla, C. D., Borrajo, G. J. C., et al. (2024). *Current status of newborn bloodspot screening worldwide 2024: A comprehensive review of recent activities (2020–2023). International Journal of Neonatal Screening, 10*, 38. Link

63 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet Journal of Rare Diseases*, *20*(1). Link

64 National Health Security Office. (2024, April 22). *From 'Treats All Diseases' to 'Treatment Anywhere': The Transformation of Thailand's Universal Coverage Scheme*. NHSO. Link

Viet Nam

Viet Nam's newborn screening services have expanded gradually since 2006, when the Ministry of Health began implementing national programmes on prenatal and newborn screening, diagnosis, and treatment of selected congenital and genetic conditions. Current newborn screening focuses on:

1. Phenylketonuria
2. Congenital Hypothyroidism
3. G6PD Deficiency

These screenings are delivered primarily through heel-prick blood sampling at commune-level health stations, with district health facilities supporting prenatal screening using ultrasound techniques.[65] In December 2020, the Prime Minister approved a national programme to expand prenatal and newborn screening through to 2030, establishing a nationwide implementation framework across more than 10,000 districts and communes. Between 2016 and 2019, coverage improved, with the proportion of newborns screened rising from 23% to around 40%, although overall national coverage remains below 50%.[66] The Ministry of Health has set longer-term targets to achieve 90% newborn screening coverage within the next decade, alongside expanding service delivery points and regional screening and diagnostic centres.

Diagnostic services remain concentrated in Hanoi and Ho Chi Minh City, limiting accessibility in rural, mountainous, and ethnic minority areas, where uptake of prenatal and newborn screening continues to lag significantly behind national averages. Although screening activities have been implemented nationwide, the programme faces persistent challenges, including uneven service networks, limited awareness, and financing constraints. National health insurance only partially covers the costs of newborn screening, and families often continue to bear out-of-pocket expenses.[67] Private hospitals and companies such as Vinmec and Medlatec provide more advanced genetic testing services, but high costs restrict their use largely to higher-income populations.

65 Nguyen, T. T., Le, Q. T., Hoang, D. T., Du Nguyen, H., Ha, T. M. T., Nguyen, M. B., Ta, T. T., Tran, N. T., Trinh, T. H. N., Doan, K. P. T., Lam, D. T., Tran, S. T. T., Nguyen, T. X., Le, H. T., Ha, V. T., Nguyen, M. H., Le, B. K., Duong, M. L., Pham, T. H., Tran, A. T., ... Giang, H. (2022). Massively parallel sequencing uncovered disease-associated variant spectra of glucose-6-phosphate dehydrogenase deficiency, phenylketonuria and galactosemia in Vietnamese pregnant women. *Molecular genetics & genomic medicine, 10*(7), e1959. Link

66 Vietnam+. (2023, March 26). Prenatal, newborn screening programme helps improve population quality. *Vietnam+ (VietnamPlus)*. Link

67 VnExpress. (2023, July 7). Vietnamese people spend twice as much money on healthcare out of pocket as recommended. *vietnam.vn*. Link

- Diagnostic access for rare diseases remains a public health and equity priority in ASEAN.
- Singapore, Thailand, and the Philippines show how coordinated governance and universal coverage can expand access.
- Indonesia, Malaysia, and Viet Nam are laying important foundations through ongoing reforms.
- Continued investment, harmonised standards, and stronger regional collaboration can accelerate early detection.
- Shared data systems, pooled procurement, and capacity-building can help make early diagnosis a guaranteed right for every child in Southeast Asia.

5.2 Referral Pathways and Efforts to Reduce Delays

Across ASEAN, the length of the diagnostic journey for rare conditions is strongly shaped by how quickly patients move through the system from first presentation to confirmatory testing and specialist care. Three design features consistently shorten time to diagnosis: clearly documented referral protocols with named centres of excellence; administrative policies that remove avoidable gatekeeping; and digital tools that connect services and clinicians in real time. Countries that combine these features report fewer missed referrals and faster initiation of care. In contrast, countries that rely on paper-based approvals or fragmented pathways continue to see multi-year delays, repeat consultations, and catastrophic out-of-pocket spending while families "shop" for answers.

Indonesia

Indonesia's referral pathway for rare-disease patients is governed by the Minister of Health Regulation No. 16 of 2024 on the System of Referral for Individual Health Services, which establishes a tiered and digitally integrated mechanism linking primary, secondary, and tertiary facilities.[68] Moreover, the rare disease referral pathway begins at the primary healthcare level (puskesmas), with patients moving through a tiered system to secondary and then tertiary hospitals for specialised care under the JKN national health insurance. However, this journey is frequently challenging due to a protracted "diagnostic odyssey," as frontline clinicians often lack awareness of rare diseases, leading to misdiagnoses and delays. Access to crucial resources, such as advanced genetic testing, is limited. However, national initiatives such as the Biomedical Genome Science Initiative (BGSi) and collaborations with private labs are improving diagnostic

68 Ministry of Health Indonesia. (2024, November 12). *Peraturan Menteri Kesehatan Nomor 16 Tahun 2024 Sistem Rujukan Pelayanan Kesehatan Perseorangan*. JDIH BPK. Link

capabilities.[69] Despite progress, challenges persist with healthcare provider knowledge, geographic access to specialised centres, and financial burdens. However, the government and NGOs are increasingly focused on improving healthcare equity and referral services for complex conditions, including rare diseases.

Once the patient is stabilised or diagnosed, they might be referred back to lower-level facilities for continued treatment or rehabilitation under specialist supervision, maintaining continuity within the JKN national insurance system.[70] The Minister of Health Regulation No. 16 of 2024 also mandates integration with the National Health Information System (SIKNAS) to enable real-time monitoring of facility capacity and specialist availability, ensuring efficient, data-driven referrals. For rare diseases, horizontal referrals between hospitals of the same level are permitted when specific diagnostic or laboratory capabilities are unavailable.

Malaysia

Malaysia's referral pathways for rare diseases are being strengthened through the NPRD (2025), which designates Hospital Kuala Lumpur (HKL) as the national referral hub and Hospital Pulau Pinang, and academic medical centres such as Pusat Perubatan Universiti Malaya (PPUM), Hospital Pakar USM Kubang Kerian Kelantan (HUSM), Hospital Pakar Kanak-Kanak UKM (HPKK UKM), and several other private hospitals.[71] These facilities anchor a coordinated system linking diagnosis, treatment, and follow-up care. The Cluster Hospital Initiative further improves efficiency by connecting tertiary and district hospitals, ensuring continuity of care closer to patients' homes. Despite these advances, Malaysia still faces major challenges. There is no comprehensive national registry or epidemiological database, regulatory processes for orphan drugs remain slow, and a severe shortage of geneticists, counsellors, and laboratory professionals continues to delay accurate diagnosis.[72]

The Philippines

The Philippines has institutionalised a cradle-to-clinic referral pathway that links screening, confirmation, and long-term care. The Newborn Screening Reference Centre (NSRC) operates a national system in which positive results

69 Ministry of Health Indonesia. (2022, August 14). *Mengenal Biomedical Genome Science Initiative (BGSI)*. Kemenkes. Link
70 RS PKU Muh Surakarta. (2022, June 9). *Programme rujuk balik pasien kronis*. rspkusolo.com. Link
71 Ministry of Health Malaysia. (2025). *National Policy for Rare Diseases in Malaysia* (MOH/P/PAK/576.25(GU)-e). Medical Development Division. Link
72 Malay Mail. (2025, September 11). Malaysia's path to becoming Asean's hub for rare disease healthcare – Spinal Muscular Atrophy Malaysia. *Malay Mail*. Link

from Newborn Screening Centres (NSCs) are referred to Newborn Screening Continuity Clinics (NBSCCs) for confirmatory testing, subspecialist referral, and life-long follow-up. The chain of accountability is explicit: NSCs conduct short-term follow-up and hand off confirmed cases to NBSCCs; Department of Health regional offices support monitoring and patient navigation. This architecture reduces leakage between screening and care and has become the backbone for rapid referrals in metabolic and endocrine conditions. To mitigate specialist shortages outside major hubs, the country has also relied on teleradiology and other telehealth tools to expedite specialist reads and decision-making, reducing unnecessary travel and shortening diagnostic intervals when imaging informs the differential diagnosis.[73]

Singapore

In Singapore, the referral pathway for rare diseases begins with a primary or secondary care provider referring a patient to one of the specialist genetics clinics at major public hospitals, such as KKH, National University Hospital (NUH), or Singapore General Hospital (SGH). Electronic referrals are typically processed through the National Electronic Health Record (NEHR) system. A multidisciplinary team of specialists, including clinical geneticists and counsellors, reviews the patient's case to coordinate further evaluation. Patients undergo diagnostic testing, which can include biochemical, molecular, or genomic analysis, depending on the suspected condition. Confirmed cases are discussed in a genetics case conference to finalise the diagnosis and treatment plan. Following genetic counselling for the family, confirmed patients are referred to disease-specific clinics for long-term clinical management. Throughout this process, data are integrated with institutional databases and linked to research networks such as the SGRDP Registry and the Singapore RDMM Network to support clinical advancement and research. Eligible patients at public healthcare institutions can seek assistance with high-cost treatments through medical social workers, who facilitate applications for various financial schemes, including the RDF.[74,75,76]

Thailand

Thailand's referral pathway for rare-disease patients operates within the UCS, which is managed by the NHSO. Patients typically begin their journey at local hospitals or community clinics, where a physician identifies signs of

73 Department of Health Philippines. (2023). *Manual of Operations: National Comprehensive Newborn Screening System*. Department of Health. Link
74 SingHealth. (n.d.). *Singapore Rare Disease Models and Mechanisms (RDMM) Network - Genomic Medicine Centre*. Link
75 KK Women's and Children's Hospital. (n.d.). *Genetics*. Link
76 National University Polyclinics. (n.d.). *Be Referred to a Specialist Outpatient Clinic (SOC)*. NUP. Link

a possible rare or inherited condition. When suspicion of 24 rare metabolic disorder with emergent condition arises, the local hospital can use the fast-track consultation and referral to 7 rare disease service centres, by-passing the regular sequential referral. As for the other rare disorders, the cases are referred through the UCS system to one of seven designated tertiary referral hospitals with clinical genetics and metabolic expertise, located mainly in Bangkok and Khon Kaen. These include Chulalongkorn Hospital, Siriraj Hospital, Ramathibodi Hospital, Thammasat University Hospital, Queen Sirikit National Institute of Child Health, Phramongkutklao Hospital, and Srinagarind Hospital. At these centres, multidisciplinary teams composed of geneticists, metabolic specialists, paediatricians, and laboratory scientists conduct biochemical and genetic investigations.[77,78] Additionally, a number of patients with rare disorders get genomic testing and are diagnosed through the national research initiative, Genomics Thailand, during 2020-2024, using the Thailand Rare and Undiagnosed Disease Network (T-RUN), which follow the 7 rare disease service networks.

The referral process is governed by NHSO policy, which specifies that patients covered under UCS for one of the 24 recognised rare or inherited metabolic diseases are eligible for full diagnostic and treatment benefits at these centres. Once diagnosis and treatment plans are confirmed through case conferences, patients remain under the supervision of tertiary hospitals for clinical management and long-term monitoring. The 2019 NHSO guidance on rare-disease management outlines the need for coordination between tertiary centres and local hospitals to ensure continuity of care and data reporting. Telemedicine is increasingly used across Thai hospitals for general specialist consultations and follow-up services, but it has not yet been institutionalised within the rare-disease referral pathway.[79] Non-invasive prenatal testing (NIPT) has been available in Thailand since 2012, offered on a self-pay basis. As of March 2025, the Thai NHSO (UHC) has launched non-invasive prenatal testing (NIPT) for all pregnant women in Thailand, although the service system is still facing some challenges[80].

77 The Nation. (2021, March 12). 24 rare diseases included in universal coverage, but challenges remain. *Nationthailand*. Link

78 National Health Security Office. (2023, September 5). *Tandem mass spectrometry newborn screening is available nationwide*. NHSO. Link

79 Gaewkhiew, P., Kittiratchakool, N., Suwanpanich, C., Saeraneesopon, T., Athibodee, T., Kumluang, S., Chuanchaiyakul, T., Liu, S., Chanpanitkitchot, S., Laosuangkul, A., & Isaranuwatchai, W. (2024). Telemedicine utilization in tertiary, specialised, and secondary hospitals in Thailand. *Telemedicine Reports*, *5*(1), 237–246. Link

80 National Health Security Office. (2025). Link

Viet Nam

A significant reform in Viet Nam's health insurance system took effect on 1 January 2025, fundamentally reshaping how patients with rare and severe diseases access specialised care. Under the amended Health Insurance Law (Law No. 51/2024/QH15) and the Ministry of Health's Circular 01/2025/TT-BYT,[81] patients diagnosed with 62 designated rare or severe diseases, including metabolic disorders, advanced heart failure, organ transplants, and malignant tumours, are now entitled to full insurance coverage for medical examination and treatment at any public hospital without referral documents. This reform, which expanded the previous list by 20 conditions, eliminates the long-standing requirement for patients to obtain multiple referral letters before accessing central or provincial hospitals. It allows direct treatment at specialised and high-tech facilities such as the National Cancer Hospital (K Hospital), where more than 20,000 insured cancer patients received care in the first days of the new regulation. By removing bureaucratic barriers and guaranteeing 100% coverage, the reform represents a significant step toward equitable access, faster diagnosis, and improved outcomes for people living with rare and severe diseases in Viet Nam.[82]

- Referral pathways for rare diseases in several ASEAN countries remain uneven due to systemic and operational gaps.
- Structured systems and digital tools exist but many patients still face long diagnostic delays.
- Limited specialists, inconsistent protocols, and weak primary-tertiary coordination hinder timely referrals.
- Paper-based processes, out-of-pocket costs, and low provider awareness add further barriers.
- Stronger workforce capacity, institutional systems, and equitable financing are needed for effective, timely diagnosis.

5.3 Genomic Capacity and Data-Sharing Mechanisms

Genomic testing should not be regarded as a universal confirmatory step following newborn screening. Newborn screening programmes are designed as population-level early detection systems that identify infants at increased risk of specific conditions, rather than as diagnostic tools in themselves. For many screened disorders, diagnostic confirmation continues to rely primarily

81 Ministry of Health Viet Nam. (2019). *Circular No. 26/2019/TT-BYT on List of Orphan Drugs*. Socialist Republic of Viet Nam. Link

82 The Ministry of Finance Viet Nam. (2024, November 28). *Amendments to health insurance law ensure benefits of card holders*. Vietnam Social Security. Link

on established biochemical investigations and clinical assessment, which are often more immediately informative for clinical decision-making and treatment initiation. In this context, genomic testing typically serves a complementary function, for example to clarify equivocal findings, support differential diagnosis where multiple genetic aetiologies are possible, or provide molecular characterisation that may inform prognosis, long-term management, or family counselling. Its role therefore varies by condition, clinical context, and system capacity, rather than constituting a routine or automatic step for all screen-positive cases.[83,84]

In the context of symptom-led diagnoses, which remain the predominant pathway for many individuals with rare diseases, genomics can play a more substantive role in supporting earlier and more accurate diagnosis, particularly where appropriate specialist expertise, laboratory capacity, and clearly defined clinical pathways are in place. When effectively integrated into multidisciplinary care, genomic testing has the potential to reduce prolonged and fragmented diagnostic journeys, limit unnecessary investigations, and facilitate more timely access to appropriate services.

Indonesia

Indonesia's genomic capacity has moved from disparate pilots to a networked platform anchored by the Ministry of Health's BGSi and complemented by BRIN facilities. BGSi is standing up a distributed biobank ecosystem with a single database layer and seven national referral hospitals aligned by clinical domain: Persahabatan for respiratory disease, Dharmais for cancer, National Brain Centre PON for neurology, Sulianti Saroso for infectious disease, Cipto Mangunkusumo (RSCM) for diabetes, Sardjito for rare diseases, and RSUP Prof. Ngoerah for health and beauty; public-private collaboration includes NGI (Naleya Genomics Indonesia), Bumame, and BGI (Beijing Genomics Institute), together with an MoU on thalassaemia prevention and a National Gene Bank initiative.[85]

Operational capacity expanded during and after COVID-19: UNDP and MoH equipped 17 whole-genome sequencing labs with plans to reach 29, while Sam Ratulangi University's node has produced multiple WGS batches and received competency certification; in parallel, BRIN's Sequencing Lab runs

83 Yu, B., Yang, Y., Zhou, L., & Wang, Q. (2024). Evaluating a novel newborn screening methodology: combined genetic and biochemical screenings. *Archives of Medical Research, 55*(2), 102959. Link

84 Carli, D., Quarello, P., Porta, F., Cagnazzo, C., Zucchetti, G., Proto, C. F., Gianasso, R., Biamino, E., Carbonara, C., Coscia, A., Parlato, C., Fenoglio, B., Guarrera, S., Spada, M., Mussa, A., Minucci, S., & Fagioli, F. (2025). A Genomic Sequencing Approach to Newborn Mass Screening and Its Opportunities. *JAMA network open, 8*(10), e2538198. Link

85 Nasution, R. (2023, February 20). Ministry builds collaboration for biobank ecosystem. *Antara News*. Link

Illumina, MGI, and Oxford Nanopore platforms with defined access pathways to channel sequence data into a national repository.[86,87] Policy support has also improved. Indonesia's 2023 Health Law recognises genomics as a key biomedical technology. It sets rules for biobanks, requiring specimens and data to be kept within the country, and managing cross-border transfers through material transfer agreements. However, the law does not yet clearly define how pathogen genomics should be used in routine surveillance.[88] Strategic debates continue around cost and prioritisation. Commentaries note a two-year goal to collect 10,000 Indonesian genomes, supported by mixed funding. They also warn that long-term sustainability, the growth of the bioinformatics and clinical-genetics workforce, and fair distribution of benefits must be ensured to prevent resource waste and protect privacy.[89]

Data-sharing mechanisms sit at the intersection of sovereignty, privacy, and global health security. Indonesia championed rapid genomic data-sharing during its G20 presidency and cites GISAID as a workable model. By April 2021, GISAID housed 1.2 million SARS-CoV-2 sequences, and by April 2022, it had surpassed 10 million. Yet, Indonesia's contribution was only about 10,000 to 20,000 entries, highlighting a performance gap that policy can close through funded deposition targets, ethics governance, and secure pipelines from BGSi and BRIN labs.[90] The legal basis for protecting human genomic data is now clearer under Law No. 27 of 2022 on Personal Data Protection. The law classifies genetic and health information as specific data and requires controllers to prove consent, maintain confidentiality, and prevent unlawful access. These provisions apply directly to biobanks and clinical genomics operated by public and private entities, as outlined in Articles 20 to 50, with genetic data listed in Article 4.[91] Still, the literature highlights significant gaps for genomics. These include more explicit rules for secondary use and de-identification of data, stronger safeguards to prevent genetic discrimination in insurance and employment, clear guidelines for how public bodies handle genomic data, and practical enforcement that focuses mainly on corrective administrative action.

86 Tamara, M. (2025, February 5). *Eastern Indonesia Advances Healthcare with UNDP-supported Bioinformatics Initiative*. UNDP. Link
87 BRIN Public Relations. (2024, June 9). *BRIN Sequencing Lab Supports Research Based on Genomic Data*. BRIN. Link
88 Tundang, R. E. (2023, August 29). Genomic data, privacy and equity in Health Law. *The Jakarta Post*. Link
89 Fletcher, L. (2023, May 16). *World of Genomics: Indonesia*. Front Line Genomics. Link
90 Assegaf, F. (2022, April 30). Genome data-sharing to address future pandemic potentials. *Antara News*. Link
91 Syailendra, M. R., Lie, G., & Sudiro, A. (2024). Personal Data Protection Law in Indonesia: Challenges and Opportunities. *Indonesia Law Review*, *4*, 56–72. Link

Malaysia

Malaysia's genomic capacity is progressing rapidly from fragmented research initiatives to a coordinated, nationally integrated platform supporting clinical care and precision medicine. The country's flagship MyGenom Project, jointly led by the Ministry of Science, Technology and Innovation (MOSTI) and the MoH, aims to sequence 10,000 genomes to establish a Malaysian reference genome and characterise genetic variation across the country's major ethnic groups. Its first phase (2024-2025) will complete 2,400 genomes, with standardised procedures for DNA extraction, quality control, and sequencing under the leadership of the Malaysia Genome & Vaccine Institute (MGVI) and the Institute for Medical Research (IMR).[92] The initiative builds on the Academy of Sciences Malaysia's Precision Medicine Task Force and earlier work by the Malaysian Node of the Human Variome Project (MyHVP), which developed national variant databases aligned with global nomenclature standards to improve variant interpretation.[93]

Data sharing mechanisms are being developed to ensure that genomic advances are implemented ethically and sustainably. The MyGenom Project database will serve as a centralised national repository enabling controlled access for clinical, research, and diagnostic use, while adhering to Malaysia's Guidelines on Ethical Issues in the Provision of Medical Genetics Services and Guidelines on the Use of Human Biological Samples for Research, which set standards for informed consent, confidentiality, and secondary data use.[94] Malaysia has also demonstrated regional leadership in pathogen genomics. During the COVID-19 pandemic, a consortium of seven to ten laboratories under MOSTI and MoH conducted nationwide genomic surveillance, contributing sequences to global databases such as GISAID, and rapidly identifying Beta, Delta, and Omicron variants to inform public health policy.[95] According to a 2024 Nature Microbiology regional analysis, Malaysia ranks among the top four Southeast Asian countries with advanced genomic surveillance systems, routinely uploading over 75% of pathogen sequences to public repositories and engaging policymakers in 77% of genomic data-sharing cases.[96] Moving forward, Malaysia's challenge is to translate these data-sharing practices

92 BERNAMA. (2024, August 29). *MOSTI's MyGenom project targeting collection of 10,000 genome sequence for local genetic references*. Link

93 Nik Hassan, N. N., Plazzer, J.-P., Smith, T. D., Halim-Fikri, H., Macrae, F., Zubaidi AL, A., & Zilfalil, B. A. (2016). Harmonizing the interpretation of genetic variants across the world: the Malaysian experience. *BMC Research Notes, 9*(1). Link

94 MalaysiaKini. (2024, September 2). Launching of the MyGenom Project: Advancing Precision Medicine and Genomic Research in Malaysia. *MalaysiaKini*. Link

95 UKM Medical Molecular Biology Institute. (2022, October 7). *COVID-19 Genomic surveillance in Malaysia: challenges and opportunities*. UKM. Link

96 Getchell, M., Wulandari, S., de Alwis, R., Agoramurthy, S., Khoo, Y. K., Mak, T.-M., Moe, L., Stona, A.-C., Pang, J., Momin, M. H. F. H. A., Amir, A., Andalucia, L. R., Azzam, G., Chin, S., Chookajorn, T., Arunkumar, G., Hung, D. T., Ikram, A., … Jha, R. (2024). Pathogen genomic surveillance status among lower resource settings in Asia. *Nature Microbiology, 9*(10), 2738–2747. Link

into national law, strengthen laboratory accreditation and bioinformatics infrastructure, and secure sustainable financing for genomics programmes so that pandemic-era progress becomes a lasting foundation for precision medicine in rare diseases.

The Philippines

The Philippines has established a growing national infrastructure for genomics, integrating research, clinical applications, and public health uses. The Philippine Genome Centre (PGC), a multidisciplinary research and service unit under the University of the Philippines, provides sequencing, bioinformatics, and biobanking services that support research in health, agriculture, biodiversity, and forensics. Its 2019-2025 Strategic Plan identifies priorities in social responsibility, operational efficiency, and nationwide linkages through satellite facilities in Visayas and Mindanao.[97] The Department of Science and Technology (DOST), through the Philippine Council for Health Research and Development (PCHRD), has been a key funder of health genomics research, allocating over 320 million pesos in 2022 to develop diagnostics, therapeutics, and precision medicine applications.[98] Research programmes such as the Philippine Genomes Centre and studies on X-linked Dystonia Parkinsonism (XDP) have expanded local understanding of genetic variations relevant to Filipino populations.[99] The Institute of Human Genetics at the University of the Philippines–National Institutes of Health complements this capacity through clinical evaluation, molecular diagnostics, biochemical genetics, and genetic counselling services, supported by ISO 9001:2015-certified laboratories.[100] These institutional investments collectively form the technical and scientific foundation for the country's genomics and genetic medicine initiatives.

Data-sharing and genomic surveillance are integrated into several national systems. The Department of Health's Antimicrobial Resistance Surveillance Programme (ARSP), operating since 1988 across 24 sentinel sites, incorporated WGS in 2018 through a collaboration between the Research Institute for Tropical Medicine (RITM), the Centre for Genomic Pathogen Surveillance (CGPS), and the University of Oxford's Big Data Institute.[101] Local sequencing

97 Philippine Genome Centre. (n.d.). *Genomics for a better Philippines: Strategic Plan 2019-2025*. University of the Philippines. Link

98 Philippine Council for Health Research and Development. (n.d.). *Research on genomics significantly contributed to PH healthcare - DOST Secretary*. Link

99 Padilla, C. D., & Cutiongco-de la Paz, E. M. (2016). Genetics and genomic medicine in the Philippines. *Molecular genetics & genomic medicine*, *4*(5), 494-503. Link

100 Collins, A. (2024, February 20). *World of Genomics: The Philippines*. Front Line Genomics. Link

101 Argimón, S., Masim, M. A. L., Gayeta, J. M., Lagrada, M. L., Macaranas, P. K. V., Cohen, V., Limas, M. T., Espiritu, H. O., Palarca, J. C., Chilam, J., Jamoralin, M. C., Jr., Villamin, A. S., Borlasa, J. B., Olorosa, A. M., Hernandez, L. F. T., Boehme, K. D., Jeffrey, B., Abudahab, K., Hufano, C. M., … Carlos, C. C. (2020). Integrating whole-genome sequencing within the National Antimicrobial Resistance Surveillance Programme in the Philippines. *Nature Communications, 11*(1). Link

using Illumina MiSeq and a dedicated bioinformatics server enables analysis and interpretation of genomic data within the country. Findings are disseminated through national reports and interactive online platforms such as Microreact and Pathogenwatch, which link genomic, epidemiological, and resistance data to identify high-risk clones and inform infection control.[102] The Philippines contributes AMR genomic data to the World Health Organization's Global Antimicrobial Resistance Surveillance System (GLASS), reflecting growing national ownership in genomic data generation and use. The World Health Organization's regional review of genomics in the Western Pacific notes the Philippines among countries advancing genomics integration through institutional capacity building, local sequencing, and international collaboration.

Singapore

Singapore's genomic capacity has matured into one of the most advanced and clinically integrated systems in Asia, combining next-generation sequencing (NGS) technologies with a coordinated national precision medicine strategy. The Genetics Service at KK Women's and KKH manage approximately 2,000 patients annually, including around 500 new referrals each year, and is supported by over two decades of institutional experience in clinical genetics.[103] Flagship BRIDGES programme.[104,105] In selected cohorts using trio or quad sequencing, yields exceeded 50%, demonstrating the advantage of family-based genomic analysis.[106] The RapidSeq Initiative, established in 2018, extends this capacity to critically ill patients in the neonatal or paediatric intensive care units, achieving a 40% diagnostic rate within 10-14 working days during its pilot phase.[107] Such speed and precision are clinically meaningful: among patients who received a confirmed diagnosis through BRIDGES, 100% experienced changes in genetic counselling and about 27% had adjustments in clinical management, including curative interventions such as bone marrow transplantation.[108] Downstream, the SingHealth Duke-NUS Genomic Medicine Centre (SDGMC) ensures consistent and multidisciplinary care across institutions, integrating genetics clinics into routine hospital services to reduce post-diagnosis fragmentation, and support life-long management.

Singapore's data sharing mechanisms complement these clinical initiatives through secure, federated, and diversity-driven systems that link local data

102 Ibid.
103 SingHealth. (n.d.). *Clinical Overview and objectives*. SingHealth DukeNUS. Link
104 Ibid.
105 Precision Health Research. (2021, July 21). *Ending the rare disease odyssey*. PRECISE. Link
106 Precision Health Research. (2021, June 23). *Bridging the Diagnostic Gap*. PRECISE. Link
107 Kam, S., Ling, K. A., & Ying, L. J. (2020, December 22). *Hope Should Not Be Rare for Patients with Rare Diseases*. SingHealth Duke-NUS Genomic Medicine Centre. Link
108 Precision Health Research. (2021, June 23). *Bridging the Diagnostic Gap*. PRECISE. Link

sovereignty with global research collaboration. As a key contributor to the federated Genome Aggregation Database (gnomAD), Singapore provides variant frequency data across its multi-ethnic population – Chinese, Malay, and Indian – while retaining all individual-level data within national borders to protect privacy.[109] Large-scale genomic initiatives such as SG10K Health and SG100K have sequenced up to 100,000 individuals and identified high-prevalence pathogenic variants, including those linked to familial hypercholesterolaemia (1 in 140) and hereditary breast and ovarian cancer (1 in 150).[110] These datasets improve the accuracy of variant interpretation and support precision health efforts across Southeast Asia, particularly given that 61% of variants identified in Singapore's multi-ethnic cohort were previously unknown.[111]

The Undiagnosed Disease Programme (UDP) and the Rare Disease – Models and Mechanisms (RDMM) network support discovery by linking clinicians with laboratory scientists to confirm new disease genes using model organisms, while using international standards such as the Human Phenotype Ontology and GA4GH Phenopackets to ensure data can be shared and interpreted consistently.[112] Singapore's contributions to global resources such as ClinVar, GeneMatcher, and Matchmaker Exchange help address the long-standing under-representation of Asian populations in genetic databases, a gap that previously left more than 60% of Asian variants uncatalogued.[113] Collectively, these efforts demonstrate a robust, privacy-preserving infrastructure that accelerates rare disease diagnosis, improves equity for underrepresented populations, and establishes a durable foundation for regional precision medicine collaboration.

Thailand

Thailand's genomic capacity has been strategically developed through the Genomics Thailand Initiative, a national precision medicine programme launched in 2019 and operationalised under a five-year Integrated Action Plan (2020–2024) with a budget of THB 4,570 million (USD 140 million). The initiative aims to sequence 50,000 Thai genomes and expand precision medicine beyond oncology to include rare and undiagnosed diseases (RUDs), noncommunicable diseases, infectious diseases, and pharmacogenomics.[114,115] Core infrastructure under the National Science and Technology Development Agency includes

109 Precision Health Research. (2025, May 14). *Decoding Diversity with gnomAD*. PRECISE. Link
110 Ibid.
111 Precision Health Research. (2021, June 23). *Bridging the Diagnostic Gap*. PRECISE. Link
112 Groza, T., & Vasilevsky, N. (2024, June 25). *Uncovering and overcoming common data-sharing challenges in the Rare Disease landscape*. Global Alliances for Genomics & Health. Link
113 Precision Health Research. (2021, June 23). *Bridging the Diagnostic Gap*. PRECISE. Link
114 National Science and Technology Development Agency. (2020, July 13). *Genomic medicine in Thailand*. NSTDA Eng. Link
115 Thailand Centre of Excellence for Life Sciences. (n.d.). *Genomics Thailand Strategic Roadmap*. Link

the National Biobank of Thailand, which operates the Genome Data Bank and manages GeTH-50K data access and security; the National Omics Centre for sequencing and multi-omics services; and high-performance compute capacity to process large-scale whole-genome datasets.[116] Early findings show that Thai-specific whole-genome datasets are now helping improve variant interpretation and pharmacogenomics, signalling a shift from research pilots to clinical use. At the same time, experts stress the need to maintain investment in bioinformatics pipelines, interoperable data systems, and a larger analyst and clinical-genomics workforce to support nationwide implementation.

Thailand's data-sharing mechanisms combine national stewardship with international collaboration. The National Biobank of Thailand functions as the principal data steward, operating a controlled-access architecture that enforces authentication, de-identification, and audit trails across collaborating institutions. To strengthen cross-institutional collaboration, the government has prioritised harmonising data formats across hospitals, universities, and laboratories to address longstanding fragmentation in database ownership. Alongside national efforts, the 2023 UK-Thailand Memorandum of Understanding connects Genomics Thailand's 50,000-genome project with the UK's 100,000 Genomes Project, focusing on rare and undiagnosed diseases and cancer, and creating technical working groups for data handling, training, and joint research.[117] In parallel, Thailand's Open Data for Health Policy Initiative – implemented through data.go.th under the Digital Government Development Agency and supported by WHO and ThaiHealth – applies open-access principles to de-identified public-health datasets, promoting transparency while protecting individual privacy.[118]

Viet Nam

Viet Nam's genomic capacity has expanded rapidly from limited beginnings to a more coordinated, clinically relevant ecosystem. Until recently, Viet Nam was severely under-represented in global datasets, with only 99 Vietnamese genomes included in the 1000 Genomes Project, which constrained variant interpretation and clinical application.[119] This has changed through sustained investment in both clinical and research infrastructure. Non-invasive prenatal testing (NIPT), introduced in 2007, is now widely available for detecting trisomies 21, 18, and 13 with over 99% accuracy.[120] Thalassaemia remains a

116 National Biobank of Thailand. (n.d.). *About us*. National Biobank. Link
117 Department of Health and Social Care, & Markham, Lord. (2023, January 20). *UK landmark genomic partnership with Thailand to unite against health threats*. Gov.UK. Link
118 Health Intervention and Technology Assessment Programme Foundation. (2023). Unveiling Thailand's Path to Open Data for Health Policy. In *HITAP* (Issue#29). Link
119 Ormiston, S. (2023, November 28). *World of Genomics: Vietnam*. Front Line Genomics. Link
120 Ibid.

major genetic public health issue, with more than 12 million carriers and rates between 1.5% and 25% among ethnic minorities. A 2021 pilot using next-generation sequencing (NGS) is screening pregnant women and couples, marking a turning point in preventive genetics. By 2022, 66 gene sequencers had been installed nationwide, and 50 hospitals were piloting non-invasive genomic testing for early cancer detection, supported by the opening of Southeast Asia's largest sequencing centre in Hanoi.[121,122] The most significant national milestone is Vingroup's genome project, which sequenced over 1,000 Vietnamese genomes and identified more than 40 million genetic variants, resulting in the establishment of the MASH Portal for secure data access and analysis.[123] This effort was expanded by the VN1K project, which provides 1,011 high-depth genomes, multi-omics data, and pharmacogenomic data, along with a Vietnam-specific imputation panel for improved precision medicine.[124]

Viet Nam's data-sharing ecosystem is evolving from project-based releases to standardised, legally governed platforms. The MASH Portal now provides controlled access to an extensive variant database aligned with NIH data standards. It supports rapid whole-genome analysis, enabling both national and international researchers to use Vietnamese-specific allele frequency data for clinical interpretation.[125] The VN1K dataset strengthens this system by leveraging a function-driven, graph-based framework that integrates genome, methylome, and immunogenetic data to build a local reference and reduce variant-identification errors. Vietnam's 2024 Data Law adds a formal governance structure for the entire data lifecycle, covering collection, processing, storage, and cross-border transfer.[126] It requires licensing, risk management, and stricter oversight for data classified as core or important, which includes genomic data. The law highlights the need for privacy-by-design systems, secure storage, traceable access, and strong compliance mechanisms for institutions and companies that handle sensitive biological information.

121 Kiet, A. (2021, October 9). Southeast Asia's largest-scale genome sequencing centre set up in Vietnam. *Southeast Asia's Largest-scale Genome Sequencing Centre Set up in Vietnam*. Link

122 Van, T. (2022, October 12). *Illumina and Gene Solutions cooperate to revolutionise Vietnamese healthcare*. Vietnam Investment Review - VIR. Link

123 Viet Nam News. (2021, December 16). *Vingroup completes first genome project for Vietnamese people*. vietnamnews.vn. Link

124 Tran, T. T. H., Hoang, T. H., Tran, M. H., Nguyen, N. T., Nguyen, D. T., Pham, T. M., Nguyen, N. N., Vu, G. M., Duong, V. C., Vu, Q. T., Nguyen, T. K., Nguyen, S. V., Vu, H. Q., Nguyen, T. M., Dang, T., Nguyen, H., Do, T., Le, C., Nguyen, H. T. T., . . . Vo, N. S. (2025). VN1K: a genome graph-based and function-driven multi-omics and phenomics resource for the Vietnamese population. *bioRxiv (Cold Spring Harbor Laboratory)*. Link

125 Viet Nam News. (2021, December 16). *Vingroup completes first genome project for Vietnamese people*. vietnamnews.vn. Link

126 Việt, X. B. T. (2024, December 17). *Vietnam enacts its First-Ever data law*. KPMG. Link

- Genomic capacity and data-sharing systems in ASEAN are shifting from fragmented initiatives toward more structured national approaches.
- Singapore and Thailand have advanced precision-medicine ecosystems with federated data platforms.
- Malaysia and the Philippines are building national repositories and ethical frameworks to connect research and clinical use.
- Viet Nam and Indonesia are expanding infrastructure and introducing data-governance laws to manage privacy and cross-border flows.
- Regional progress points to emerging bioinformatics networks, standardised protocols, and interoperable databases for rare-disease diagnosis and surveillance.
- Workforce limitations, financing gaps, and uneven access may widen disparities across countries.
- Ethical data-sharing standards, linked biobanks, and joint training will be essential to ensure genomic advances benefit all populations.

5.4 Centralised Rare Disease Registry

A centralised rare disease registry is a foundational component of effective rare disease policy, enabling countries to quantify disease burden, support early diagnosis, guide service planning, facilitate research, and inform financing and access decisions. Across ASEAN, however, approaches to rare disease registries remain uneven, ranging from emerging genomics-driven databases and hospital-based systems to legally mandated but still-developing national registry frameworks. The following country snapshots illustrate the current status of rare disease registry development in selected ASEAN Member States, highlighting both promising initiatives and persistent gaps in comprehensive, interoperable, and population-level rare disease data systems.

Indonesia

At present, Indonesia does not yet operate a single national rare disease registry, but a decentralised registry ecosystem is emerging, anchored by government-led genomics initiatives and major referral hospitals. A central pillar is the BGSi, which integrates whole-genome sequencing into health services and research across national referral hospitals, including RSCM and other vertical hospitals. Through this programme, Indonesia has collected genomic data from approximately 9,000 individuals, aiming to analyse around 10,000 human genomes to support disease mapping, diagnostics, and precision medicine, including for genetic and rare disorders.[127]

127 Arlinta, D. (2024, September 12). *9,000 Indonesian human genomes collected, priority for medical research*. Kompas.id. Link

In parallel, rare disease case data are maintained through tertiary centres and community-based registries. Referral hospitals such as RSCM track metabolic and genetic conditions, including Mucopolysaccharidosis and Gaucher disease,[128] to support clinical management and access to therapy. Patient-led organisations, including Indonesia Rare Disorders (IRD), complement these efforts by documenting cases and connecting families nationwide.[129] The government's rollout of Satu Sehat as the national health data platform, alongside emerging genomic systems such as Satu DNA, is intended to strengthen interoperability between clinical records and genomic data, laying the groundwork for a more unified rare disease data infrastructure.[130]

Malaysia

Currently, Malaysia does not yet have a national rare disease registry, and rare disease data remain fragmented across individual hospitals and specialist centres. Available evidence indicates that patient information is maintained at the institutional level, limiting the country's ability to generate accurate national prevalence data or systematically plan services. A national survey conducted in public hospitals identified 1,249 patients with rare diseases, with the most frequently reported conditions including Marfan syndrome (81 patients), Prader-Willi syndrome (60), Osteogenesis Imperfecta (45), MELAS (39), and MPS II or Hunter syndrome (39).[131] The same evidence confirms that Malaysia currently has only 13 rare disease specialists and approximately a dozen medical doctors working in genetic clinics, largely concentrated in major public referral facilities.

Recognising these gaps, Malaysia's NPRD 2025 formally identifies the establishment of a national rare disease registry as a core health-system priority. In parallel, Malaysia has developed an official Malaysian Rare Disease List, overseen by the Ministry of Health and updated periodically by the National Rare Disease Committee, which serves as an essential reference point for future registry development. The list divides rare diseases into several categories, including inherited metabolic diseases, neurological and neuromuscular diseases, skin diseases, endocrine diseases, bone and connective tissue disorders, rheumatological diseases, haematological diseases, immunological diseases, rare cancers, and others.

128 Azly, E. (2019, April 2). Four rare diseases found in Indonesia. *Antara News*. Link

129 S, Miftakhul F. (2017, February 28). *Indonesia Rare Disorders, Support Group untuk Penyandang Kelainan Langka*. jawapos.com. Link

130 Ministry of Health Indonesia. (2024, September 12). *Menkes Luncurkan Portal SatuDNA sebagai Bank Data Kesehatan Berbasis Genomik*. Kemkes.go.id. Link

131 Shafie, A. A., Supian, A., Hassali, M. a. A., Ngu, L., Thong, M., Ayob, H., & Chaiyakunapruk, N. (2020). Rare disease in Malaysia: Challenges and solutions. *PLoS ONE, 15*(4), e0230850. Link

The Philippines

The Philippines does not yet have a fully centralised national rare disease registry, but the establishment of such a system is formally mandated by Republic Act No. 10747 (Rare Diseases Act of the Philippines).[132] The Act requires the Department of Health (DOH), in coordination with the National Institutes of Health (NIH), to develop and maintain a secure health information system covering rare diseases, persons diagnosed with rare diseases, and designated orphan drugs and products. The Act defines a rare disease as a disease or disorder which affects fewer than one (1) in twenty thousand (20,000) persons in the population.[133] Healthcare institutions and practitioners are required to report diagnosed cases to the registry, subject to data privacy safeguards. The registry is intended to support disease surveillance, research, programme development, and evidence-based policymaking, and to serve as a national reference for service planning and resource allocation.

In parallel, interim and disease-specific registries have been developed through the Philippine Society for Orphan Disorders (PSOD) in collaboration with the University of the Philippines–NIH Institute of Human Genetics. As of 2017, the PSOD registry had documented 319 patients across 63 rare disorders, spanning metabolic, neurological, and genetic conditions.[134] Registration also carries legal and service implications: Republic Act No. 10747 designates persons with rare diseases as Persons with Disabilities (PWDs), enabling access to government support mechanisms and social protection benefits. Registry data further inform DOH programmes and support the Philippine Health Insurance Corporation (PhilHealth) in developing benefit packages for selected rare conditions, including high-cost inherited metabolic disorders.

Singapore

Singapore does not operate a single national registry for all rare diseases; rather, it employs a multi-faceted approach involving several specialised systems that cater to different stakeholders, from patients to researchers. The primary research-oriented platform is the Singapore Rare Disease Project (SGRDP) Registry, an initiative that links human genetic data with model organism research to accelerate gene discovery and the development of therapeutic interventions.[135] This registry is primarily utilised by clinicians and scientific researchers to identify genetic causes of rare conditions. Conversely,

132 National Council on Disability Affairs. (2016, March 3). *An Act Promulgating A Comprehensive Policy in Addressing the Needs of Persons with Rare Disease – RA 10747*. Link
133 Philippine Society for Orphan Disorders. (2020, February 29). *Rare Disease Spotlight: Get to know the key provisions of Republic Act 10747 and its IRR*. psod.org.ph. Link
134 Philippine Society for Orphan Disorders. (n.d.). *Rare Diseases and the PSOD Rare Disease Registry*. Link
135 SingHealth. (n.d.). *Singapore Rare Disease Project Registry*. Link

to benefit patients and support them, the Rare Disorders Society (Singapore) (RDSS) maintains its own beneficiary registry.[136] Families must provide a formal medical diagnosis to enrol and gain access to crucial services such as financial assistance programs, community engagement events, and psychosocial support networks. The RDSS actively advocates for the eventual establishment of a formal national registry to better coordinate care and resources nationwide.

Beyond these two central systems, Singapore also manages other targeted data collection efforts. A long-established example is the National Thalassaemia Registry, created in 1992 to monitor population-level screening and support genetic counselling. Additionally, the government-backed Rare Disease Fund (RDF) maintains a confidential list of patients who meet specific clinical and financial criteria for access to high-cost, life-saving medications.[137] This fragmented but functionally specific ecosystem ensures that, while there is no single aggregated rare disease registry, targeted funding distribution is addressed through dedicated mechanisms.

Thailand

To date, Thailand does not operate a unified national rare disease registry that systematically captures all rare conditions. Instead, rare disease data are generated through a combination of targeted genomic initiatives, hospital-based databases, and disease-specific programs. A major national effort is the Genomics Thailand Initiative, launched in 2019, which aims to sequence 50,000 Thai genomes across five priority disease areas: cancer, rare diseases, non-communicable diseases, emerging infectious diseases, and pharmacogenomics. Led by a national consortium of ministries, universities, and referral hospitals, the programme is building a large-scale genomic database to support diagnosis, research, and the integration of genomic medicine into routine care. Chulalongkorn University plays a crucial role through its Centre of Excellence for Medical Genomics and its leadership of the rare disease workstream, alongside the development of referral and information-sharing platforms such as the Thailand Rare and Undiagnosed Disease Network (T-RUN) to connect hospitals and specialist centres.[138,139]

In parallel, NHSO maintains administrative patient registries linked to financing and service delivery for a defined group of conditions covered under the UCS. Since 2019, NHSO has formally identified 24 rare diseases, largely inherited

136 Rare Disorders Society Singapore. (n.d.). *Rare Disorders Society (Singapore) Beneficiary Registration Form*. RDSS. Link
137 KK Women's and Children's Hospital. (n.d.). *Rare Disease Fund*. kkh.com.sg. Link
138 Rukpium, P. (1956, October 5). *Decoding the Genes – Chula Applies Genomic Medicine to Diagnosing Rare Genetic Diseases in Thailand*. Chulalongkorn University. Link
139 Gunn, S. (2023, June 20). *World of Genomics: Thailand*. Front Line Genomics. Link

metabolic and genetic disorders, for inclusion in its benefit package, enabling the tracking of patients receiving diagnostics, treatment, and long-term care support.[140] While these datasets are critical for reimbursement and planning, they cover only a limited subset of rare conditions and are not intended to as a comprehensive national rare disease registry. Current evidence indicates that Thailand still lacks an agreed national rare disease definition and a population-wide registry framework, making it difficult to estimate the overall burden of rare diseases or consolidate clinical, genomic, and service-use data across the health system.

Viet Nam

Viet Nam is in the early stages of systematising rare disease management, with no unified national rare disease registry currently in place. According to the Ministry of Health, the country is estimated to have around six million people living with rare diseases, 58% of whom are children, highlighting the scale of the challenge. To strengthen coordination, the Ministry of Health has established a Steering Committee for Strengthening Rare Disease Management, led by the Vice Minister of Health and involving policymakers, the Viet Nam Medical Association, and clinical specialists. Current efforts focus on developing national coordination mechanisms, improving diagnosis and treatment pathways, and laying the groundwork for more structured data systems, rather than operating a comprehensive national registry.[141]

Rare disease data in Viet Nam are presently captured through hospital-based and disease-specific registries, rather than a single national platform. National and regional registries exist for selected conditions, including congenital adrenal hyperplasia, Duchenne muscular dystrophy, osteogenesis imperfecta, Turner syndrome, Pompe disease, Gaucher disease, and mucopolysaccharidoses, and are led primarily by major tertiary centres such as the National Hospital of Paediatrics and Hue Central Hospital.[142] These facility-level systems support clinical management and limited surveillance but are not yet integrated into a nationwide rare disease registry. Evidence from clinical and policy discussions continues to highlight gaps in comprehensive registration, coordinated financing, and nationwide coverage, reinforcing the need for a consolidated data framework to inform planning, early diagnosis, and access to care.

140 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet Journal of Rare Diseases, 20*(1), 258. Link

141 VietNam News. (2025, August 20). *Việt Nam faces limited access to treatment for rare diseases.* vietnamnews.vn. Link

142 Huong, N. T. M. (2016, November 16). *Definition & Challenges in Rare Disease Management*. The 2nd Rare Disease Asia Conference, Malaysia. Link

5.5 Patient Perspectives on Diagnostic and Care Journeys

Across the six ASEAN countries, patients and families living with rare diseases consistently describe their diagnostic and care journeys as prolonged, fragmented, and financially burdensome. While countries differ in the maturity of their health systems, the common experience is delayed diagnosis, limited treatment options, and the emotional toll of navigating complex, poorly coordinated systems. These shared challenges underscore systemic barriers in early detection, genetic testing, and referral pathways, often compounded by low disease awareness among both health professionals and the public.

Indonesia

Patients in Indonesia face highly fragmented care pathways with limited diagnostic infrastructure. Genetic testing is available only in a few private laboratories, and families often have to send samples abroad. Because there is no national rare disease policy, care depends heavily on family resources and advocacy groups. The *Yayasan MPS dan Penyakit Langka* Indonesia has been essential in helping families navigate bureaucracy, obtain import-tax exemptions for orphan drugs and special medical foods since 2016, and access support through fundraising and awareness efforts. Patients often report long delays before diagnosis, financial strain, and emotional stress, but they also describe strong resilience supported by peer networks.[143,144]

Malaysia

The 2025 launch of Malaysia's NPPRD is an essential milestone toward a more coordinated system, but its impact will depend on addressing long-standing gaps. Before the policy, access to care was uneven, with most diagnostic and treatment services concentrated in a few major centres such as university hospitals and Hospital Kuala Lumpur, working with the Institute for Medical Research. National data show that earlier surveys identified only 1,249 patients in public hospitals, and only about 60% received the medicines or supplements they needed, highlighting limited reach and ongoing fragmentation in services and registries. Continued monitoring and strong advocacy from groups like the Malaysian Rare Disorders Society will be essential to turn the policy into real progress on timely diagnosis, treatment, and financial protection.[145,146]

143 OTC Digest. (2018, February 20). *Harapan Peni Utami untuk Anak dengan Penyakit Langka*. Link
144 Majelis Permusyawaratan Rakyat. (2023, March 29). *Deteksi Dini Harus Ditingkatkan untuk Pencegahan Penyakit Langka di Tanah Air*. mpr.go.id. Link
145 Shafie, A. A., Supian, A., Hassali, M. a. A., Ngu, L., Thong, M., Ayob, H., & Chaiyakunapruk, N. (2020b). Rare disease in Malaysia: Challenges and solutions. *PLoS ONE*, *15*(4), e0230850. Link
146 Malaysian Rare Disorders Society. (n.d.). *Who Are We*. MRDS. Link

The Philippines

Patients with rare diseases in the Philippines often face diagnostic delays that can last several years. The Rare Disease Act of 2016 (Republic Act 10747) was intended to reduce these gaps, but it remains underfunded and unevenly implemented. Most diagnostic services are in Metro Manila, mainly at the Philippine Genome Centre and the National Institutes of Health, so rural families must travel long distances or skip testing due to the cost. The Newborn Screening Programme now covers 29 conditions, but confirmatory tests and long-term care still require significant out-of-pocket spending. Many families rely on crowdfunding or help from groups such as the Philippine Society for Orphan Disorders to access enzyme replacement therapy or genetic testing.[147,148]

Singapore

Singaporean patients benefit from the country's strong genomic capacity and well-integrated care pathways. The Undiagnosed Disease Programme and BRIDGES have reduced diagnostic timelines from years to months, identifying causative mutations in more than 40% of previously unsolved cases. Families value the system's efficiency and the close coordination between clinical and research institutions, although many still face high treatment costs and limited insurance coverage for rare therapies. Patient organisations such as the Rare Disorders Society Singapore provide emotional, informational, and financial support to affected families.[149]

Thailand

Thailand's expanded newborn screening programme and the Genomics Thailand Initiative have improved early detection for genetic and metabolic disorders. Patients benefit from the UCS, which subsidises testing and some treatments, including those for thalassaemia. However, many still face long referral chains from provincial hospitals to Bangkok-based specialists and uneven access to genetic counselling. Digital referral tools and regional screening models are helping improve equity, but advanced genomic testing and enzyme therapies remain limited outside major urban centres.[150,151]

147 Padilla, C. D., Abadingo, M. E., Munda, K. V., & Therrell, B. L. (2023). Overcoming challenges in sustaining newborn screening in low-middle-income countries: the Philippine newborn screening system. *Rare Disease and Orphan Drugs Journal*, *2*(4). Link

148 INQUIRER. (2023, March 30). *Healthcare experts: Rare disease law fund remains 'inadequate.'* INQUIRER.net. Link

149 Fong, N., Lim, J. Y., Cham, B., Kam, S., Goh, C. Y., Wei, H., Tan, Y. M., Law, H. Y., Lim, W. K., Tan, E. S., Tan, E. C., & Jamuar, S. S. (2022). Rapid exome sequencing to aid diagnostics in genetic disorders: Implementation and challenges in the Singapore context. *Annals of the Academy of Medicine Singapore*, *51*(12), 798–800. Link

150 Wichajarn, K., Sawatjui, N., Prasongdee, P., Panklin, A., Sornkayasit, K., Chungkanchana, N., Tessiri, S., Wintachai, P., Dechyotin, S., Pasomboon, C., Ratanapontee, J., Thanakitsuwan, S., & Rattanathongkom, A. (2025). The Establishment of Expanded Newborn Screening in Rural Areas of a Developing Country: A Model from Health Regions 7 and 8 in Thailand. *International journal of neonatal screening*, *11*(2), 26. Link

151 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025d). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet Journal of Rare Diseases*, *20*(1). Link

Viet Nam

In Viet Nam, diagnostic journeys are often long because genetic testing capacity is limited, and clinician awareness remains low. The Ministry of Health estimates that about six million Vietnamese live with rare diseases, many of whom experience repeated misdiagnoses and high out-of-pocket costs for confirmatory testing at major hospitals such as the National Children's Hospital. Recent national initiatives, such as the VN1K genome project, aim to improve representation and diagnostic accuracy. Families continue to depend on informal patient networks and advocacy groups for financial and psychosocial support, while calling for rare diseases to be included in social health insurance benefits.[152,153]

Table 3. Summary of Patient Journey Across Six ASEAN Countries

Country	Diagnostic Services	Referral Pathways	Genomic Capacity & Data-Sharing	Rare Disease Registry	Patient Perspectives
Indonesia	CH screening mandated but coverage very low (4.37%). Expanded screening still pilot-based. High OOP for confirmatory tests; limited specialist labs.	Tiered referral system under MoH Reg. 16/2024; delays due to low frontline awareness and limited specialists. BGSi improving access.	BGSi and BRIN expanding sequencing. Growing but uneven capacity; data governance via Personal Data Protection Law. Workforce gaps persist.	Indonesia does not yet operate a single national rare disease registry, but a decentralised registry ecosystem is emerging, anchored by government-led genomics initiatives and major referral hospitals.	Long diagnostic odysseys, repeated referrals, high OOP costs, geographic access issues.
Malaysia	NBS covers CH & G6PD; pilots for IEM via MS/MS. IMR offers comprehensive confirmatory testing for IEM. Subsidised but limited lab capacity drives some private/overseas testing. Shortage of geneticists.	HKL designated national referral centre; Cluster Hospital Initiative links tertiary/district hospitals. Delays from workforce shortages.	MyGenom Project building national reference genome. Strong pathogen genomics track record; ethics guidelines for data use.	Malaysia does not yet have a national rare disease registry, and rare disease data remain fragmented across individual hospitals and specialist centres.	Delays from limited specialists; some reliance on overseas testing; psychosocial and financial burdens.

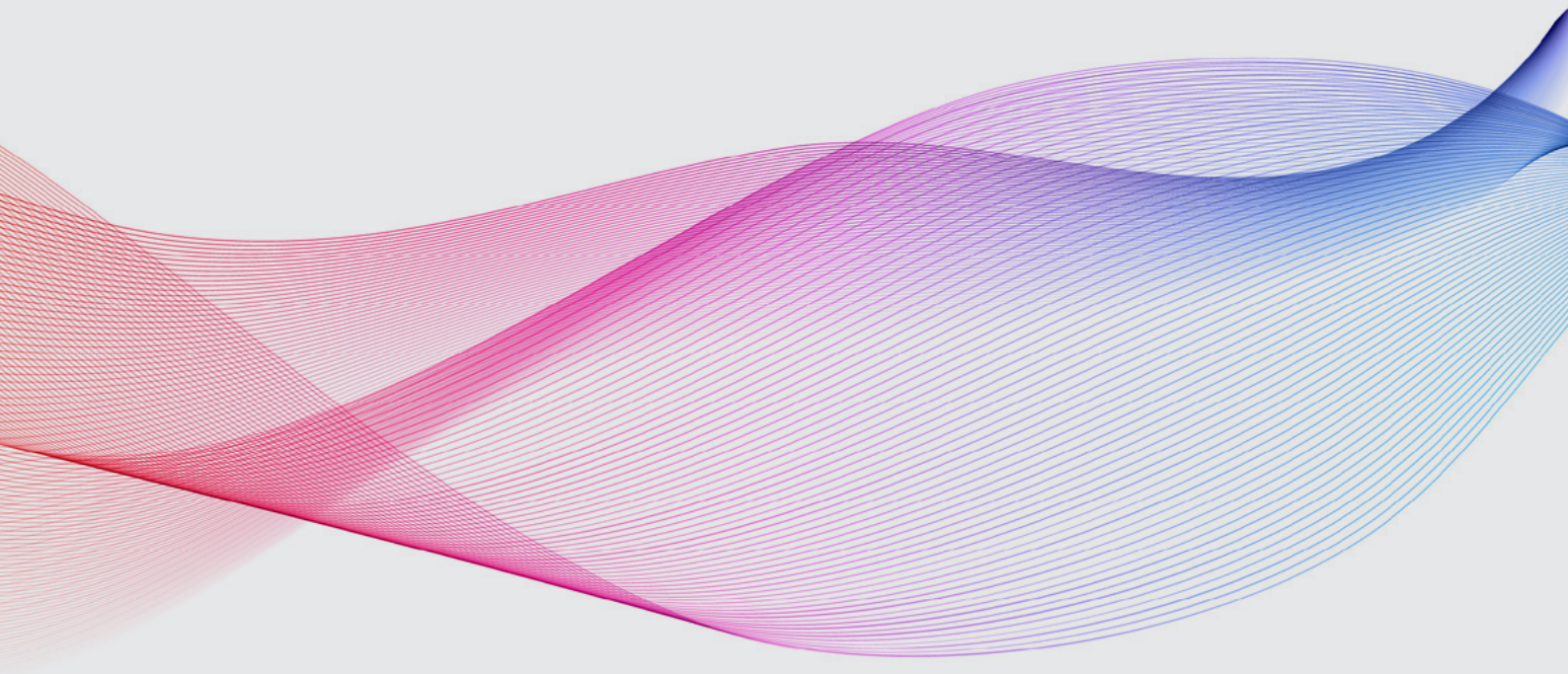
152 Vietnam+. (2023, July 28). Vietnam records about 6 million people with rare diseases. *VietnamPlus*. Link

153 Auton, A., Abecasis, G. R., Altshuler, D. M., Durbin, R. M., Abecasis, G. R., Bentley, D. R., Chakravarti, A., Clark, A. G., Donnelly, P., Eichler, E. E., Flicek, P., Gabriel, S. B., Gibbs, R. A., Green, E. D., Hurles, M. E., Knoppers, B. M., Korbel, J. O., Lander, E. S., Lee, C., . . . Abecasis, G. R. (2015). A global reference for human genetic variation. *Nature*, *526*(7571), 68–74. Link

Philippines	Expanded NBS (29 conditions) with ~92% coverage; PhilHealth covers screening but not all confirmatory tests. Access concentrated in Manila.	NBS Continuity Clinics link screening → confirmation → subspecialists. Strong national referral chain reduces leakage.	PGC, IHG-NIH, and AMR WGS network provide sequencing/bioinformatics. Strong public health genomics and national data tools.	The Philippines does not yet have a fully centralised national rare disease registry, but the establishment of a national rare disease registry system is formally mandated through Republic Act No. 10747 (Rare Diseases Act of the Philippines).	Families face financial strain for follow-up tests; access varies by region; PAGs heavily support navigation.
Singapore	Universal NBS (>40 conditions). Highly subsidised diagnostics with strong lab capacity. Advanced sequencing accessible.	NEHR-enabled electronic referrals to genetics clinics; strong multidisciplinary coordination.	SG10K/SG100K national cohorts; RapidSeq, BRIDGES, UDP. Federated data-sharing with strong privacy protection; contributes to global databases.	Singapore does not operate a single national registry for all rare diseases; rather, it employs a multi-faceted approach involving several specialized systems.	Generally faster diagnosis; advanced services reduce uncertainty; strong financial assistance options.
Thailand	UCS includes NBS and confirmatory tests; MS/MS for >40 conditions. Coverage high but confirmatory testing depends on NLEM inclusion.	UCS manages referrals to 7 tertiary centres for 24 recognised rare diseases. Strong but Bangkok-centric.	Genomics Thailand Initiative (50K genomes). National biobank, strong HPC infrastructure; UK–Thailand partnership for RUDs.	Thailand does not operate a unified national rare disease registry that systematically captures all rare conditions. Instead, rare disease data are generated through a combination of targeted genomic initiatives, hospital-based databases, and disease-specific programs.	Diagnosis faster at tertiary hospitals but travel burden significant for rural families; awareness remains low.
Viet Nam	NBS expanding but <50% national coverage. Services mainly in Hanoi and HCMC; partial reimbursement only. Private genomic testing costly.	2025 reform allows patients with 62 rare/severe diseases to access any public hospital **without referrals**; 100% insurance coverage.	Rapid growth in sequencing (66 machines). Major thalassaemia screening; Vingroup genome project expanding reference dataset. Limited national governance structures.	Viet Nam is in the early stages of systematising rare disease management, with no unified national rare disease registry currently in place.	Long travel distances; uneven access to specialists; financial hardship despite insurance reforms; stigma persists in rural areas.

CHAPTER 6.

Equitable Pathways to Treatment



CHAPTER 6.

Equitable Pathways to Treatment

6.1 Availability of Treatments

Across Southeast Asia, the availability of treatment for rare diseases reflects both progress and persistent inequities. In recent years, ASEAN countries have begun to introduce ERTs, orphan medicines products, and specialised medical nutrition into their health systems, though access remains uneven. Some, like Singapore and Thailand, have built structured mechanisms for delivering these highly specialised therapies, while others, including the Philippines, Malaysia, Viet Nam, and Indonesia, are still expanding pathways through special-access schemes, hospital initiatives, or emerging national policies. This section examines how treatments are made available across the six countries, highlighting the differences in clinical capacity, regulatory frameworks, and institutional readiness that shape the real-world accessibility of rare-disease therapies in the region.

Rare disease treatments require distinct assessment approaches because they are developed under conditions that differ markedly from those of treatments for common diseases. Clinical evidence is often based on small and heterogeneous patient populations, limited trial sites, and a lack of validated outcome measures or appropriate comparators, and frequently relies on surrogate endpoints. Given that many rare disease treatments target severe, progressive, and often life-threatening conditions with few or no therapeutic alternatives, dedicated and accelerated pathways are essential to enable timely patient access while allowing evidence to continue to evolve through post-authorisation studies, registries, and real-world data, supported by appropriate oversight and monitoring mechanisms.

Indonesia

Indonesia's access to treatment for rare diseases remains very limited. Only a handful of enzyme replacement therapies and orphan drugs are available

locally, primarily through tertiary hospitals in Jakarta and selected teaching institutions. For most other therapies, clinicians depend on the Special Access Scheme (SAS), which allows the import and controlled use of unregistered products for patients with serious or life-threatening conditions.[154] Recent regulatory updates have shortened the drug registration process for orphan and high-priority medicines. BPOM Regulation No. 23 of 2025 introduced a 100-day accelerated review pathway for medicines targeting serious and rare conditions, building on BPOM Regulation No. 24 of 2017. Yet, these have not yet translated into a noticeable increase in the number of rare-disease products in the market.

Specialised medical nutrition for metabolic disorders is available only in a few referral centres, such as Dr Cipto Mangunkusumo Hospital and Dr Sardjito Hospital, which operate limited metabolic clinics. Although Indonesia has taken steps to strengthen regulatory access, the availability of rare-disease treatments across provinces remains fragmented and inconsistent due to the concentration of specialised services in tertiary hospitals, uneven distribution of trained specialists, limited referral and follow-up capacity at sub-national levels, and the absence of a coordinated national supply and distribution mechanism. These factors underscore the need for a more integrated national strategy to ex pand clinical capacity and ensure reliable and equitable access across provinces.

Malaysia

Malaysia has made notable progress, and treatment for rare diseases is broadly available for several prioritised conditions. Enzyme replacement therapies for lysosomal storage disorders, such as Gaucher, Pompe, Fabry, and MPS I, II, and VI, are routinely administered in tertiary hospitals in accordance with the Ministry of Health's clinical guidelines.[155] Published case reports confirm ongoing treatment of these conditions within local facilities, reflecting stable supply channels and established clinical protocols.[156]

Beyond ERTs, access to other orphan medicines remains selective but expanding, guided by clinical demand and drug registration status. Specialised nutrition and metabolic formulas for IEM are available in major paediatric

154 Badan Pemeriksa Keuangan. (2024, July 23). *Peraturan Badan Pengawas Obat dan Makanan Nomor 12 Tahun 2024 tentang Perubahan atas Peraturan Badan Pengawas Obat dan Makanan Nomor 30 Tahun 2022 tentang Pemasukan Obat dan Bahan Obat melalui Mekanisme Jalur Khusus (Special Access Scheme).* JDIH BPK. Link

155 Ministry of Health Malaysia. (2012). *Guidelines for Treatment of Lysosomal Storage Diseases by Enzyme Replacement Therapy in Malaysia* (MOH/P/PAK/216.11(GU)). Link

156 Omar, A., Abdul Rahman, S., Mohamed, R., Amin Nordin, F. D., Mohamed Shakrin, N., Mukhtaroh Nasohah, S., Sallih, N. S., Abdul Azize, N. A., Abdul Wahab, S. A., Lua, S. H., Yakob, Y., Mohamed, W. A. S., Ahmad Noorden, M. S., & Abdul Jalil, J. (2025). Prevalence of lysosomal storage disease (LSD) in Malaysia. *The Malaysian journal of pathology*, *47*(2), 273–285. Link

hospitals, supported by trained metabolic teams. The 2025 National Policy for Rare Diseases marks an essential step toward a more predictable framework for ensuring consistent supply and standardised treatment protocols across hospitals.

Malaysia has also developed a national rare disease list to support policy planning, service delivery, and regulatory coordination. The list, which was last updated in March 2023, is intended to be reviewed and updated periodically by the National Rare Disease Committee under the Ministry of Health Malaysia. It currently categorises rare conditions across multiple clinical domains, including:[157]

1.	Rare inherited metabolic diseases:	123 conditions
2.	Rare neurological and neuromuscular diseases	88 conditions
3.	Rare skin diseases	23 conditions
4.	Rare endocrine diseases:	32 conditions
5.	Rare diseases affecting bone, cartilage, and connective tissue	7 conditions
6.	Rare rheumatological diseases	46 conditions
7.	Rare haematological diseases	18 conditions
8.	Rare immunological diseases	7 conditions
9.	Rare pulmonary disorders	5 conditions
10.	Rare cardiac diseases	2 conditions
11.	Rare renal diseases	56 conditions
12.	Rare gastrointestinal and hepatic diseases	5 conditions
13.	Rare malformations, developmental anomalies, and genetic syndromes	30 conditions
14.	Rare infections	2 conditions
15.	Rare cancers	44 conditions
16.	Rare eye diseases	4 conditions.

157 Ministry of Health Malaysia. (2023). *Malaysian Rare Disease List*. Link

At present, Malaysia does not have an accelerated regulatory pathway or pre-regulatory access mechanism specifically for rare disease medicines, a gap explicitly acknowledged in the 2025 National Policy for Rare Diseases. However, Malaysia has established a formal orphan medicine designation framework in accordance with the Malaysian Orphan Medicines Guideline issued by the National Pharmaceutical Services Programme (NPRA). Under this framework, a medicine must first be designated as an orphan medicine before registration, with the designation process overseen by the National Pharmaceutical Regulatory Agency through technical input from the Drug Evaluation Committee, as outlined in both the National Policy and the Drug Registration Guidance Document (Third Edition, Tenth Revision, July 2025).[158]

The Philippines

In the Philippines, treatment availability for rare diseases is partial and uneven. Several ERTs and orphan drugs are available in major tertiary hospitals, such as the Philippine General Hospital and the National Institutes of Health, which have been providing therapies for conditions like Gaucher disease, Pompe disease, and Mucopolysaccharidoses (MPS I, II, and VI) since 2017.[159,160] This focus aligns with the list of rare diseases formally acknowledged under Senate Bill No. 2279 (16th Congress of the Republic of the Philippines), which designates Gaucher disease, maple syrup urine disease, Pompe disease, galactosemia, phenylketonuria, methylmalonic acidemia, urea cycle defects, Hurler syndrome, Hunter syndrome, and Prader–Willi syndrome as rare diseases.[161]

For products not registered locally, physicians use the Department of Health's Compassionate Special Permit (CSP) mechanism, which allows the import and restricted use of unregistered drugs for patients with life-threatening conditions.[162] However, medical geneticists are available only in select hospitals, primarily those with genetic or metabolic clinics. These centres also provide biochemical testing and dietary management, but distribution beyond Metro Manila remains limited.[163] Overall, the Philippines has established clear clinical pathways and mechanisms for the treatment of rare diseases. Yet, real-world availability depends heavily on hospital-level capacity and import procedures rather than a national supply system.

158 Bahagian Regulatori Farmasi Negara (NPRA). (2025). *Drug Registration Guidance Document (DRGD) Third Edition* (No. N1-GU-01/2). Ministry of Health. Link

159 Racoma, M. J. C., Calibag, M. K. K. B., Cordero, C. P., Abacan, M. a. R., & Chiong, M. a. D. (2021). A review of the clinical outcomes in idursulfase-treated and untreated Filipino patients with mucopolysaccharidosis type II: data from the local lysosomal storage disease registry. *Orphanet Journal of Rare Diseases*, *16*(1). Link

160 Chiong, M. D., Silao, C. T., Lee, J. Y., Abarquez, C. G., & Estrada, S. C.(2009).Enzyme replacement therapy in Filipino patients with Gaucher disease and Pompe disease. *Acta Medica Philippina, 43*(1), 33-38. Link

161 Senate of the Philippines. (2014, October 6). *Senate Bill No. 2279, 16th Congress of the Republic.* Link

162 Food and Drug Administration Philippines. (2023). *Updated Guidelines for Availing Compassionate Special Permit for the Restricted Use of Unregistered or Unauthorized Drug Products including Vaccines and Medical Devices*. FDA. Link

163 Padilla, C. D., & La Paz, E. M. C. (2016). Genetics and genomic medicine in the Philippines. *Molecular Genetics & Genomic Medicine*, *4*(5), 494–503. Link

Singapore

Singapore's rare-disease treatment landscape is well-developed and well-coordinated. Most internationally recognised orphan medicines are either registered under the Health Products Act or made available through the Health Sciences Authority's Special Access Route, which permits import and use of unregistered products for specific patients when clinically justified.[164] This mechanism ensures that even therapies for ultra-rare conditions can reach patients once prescribed by accredited specialists.

Specialised nutrition for metabolic disorders is also readily available. National institutions such as KK Women's and Children's Hospital and the National University Hospital maintain dedicated biochemical genetics units that manage newborn screening, diagnosis, and dietary treatment for at least 25 rare metabolic conditions.[165,166] Singapore does not maintain a formal national list of rare diseases. Instead, access to treatment is supported through multiple mechanisms, including the Rare Disease Fund (RDF), which provides financial assistance for patients with very rare, life-threatening conditions requiring high-cost, clinically proven, life-extending medicines and currently covers 8 rare conditions, most of which have symptom onset in childhood.[167]

Thailand

Thailand offers a broad, structured range of treatments for rare diseases through its national drug and service system. Although the country does not have a single official national rare disease list, in 2020 the National Health Security Office (NHSO) introduced coverage for 24 rare diseases, primarily inherited metabolic disorders or inborn errors of metabolism, into the Universal Coverage Scheme benefits package. Furthermore, a 2025 study estimated that nearly half (46.80%) of all globally approved orphan drugs are registered in Thailand, and 22.93% are included in the NLEM.[168] Under a capitation-based reimbursement framework, the NHSO allocates approximately USD 212 per patient per year (using a 33 THB to 1 USD exchange rate) for diagnostic services and a higher tier of capitation payments; ranging from about USD 1,515 to USD 9,091 annually, to support confirmatory testing, treatment, and long-term disease management.[169] While

164 uHealth Sciences Authority. (2025, October 1). *Import and supply of unregistered therapeutic products for patient's use*. HSA. Link
165 KK Women's and Children's Hospital. (n.d.). *Biochemical Genetics and National Expanded Newborn Screening (NENS) Laboratory*. KKH. Link
166 \SingHealth. (n.d.). *Metabolic screening test for your newborn baby*. Link
167 KK Women's and Children's Hospital. (n.d.). *Rare Disease Fund*. Link
168 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025f). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet Journal of Rare Diseases*, *20*(1). Link
169 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet journal of rare diseases, 20*(1), 258. Link

Thailand's coverage for are diseases remains limited, the country stands out as one of the few in Southeast Asia with a systemised and sustainable network for the delivery of rare-disease treatment.

Viet Nam

In Viet Nam, the availability of rare-disease treatments is limited but improving. National action plans launched in 2025 recognised the country's significant unmet needs, noting that treatment access for most rare conditions remains restricted to tertiary hospitals in Hanoi and Ho Chi Minh City.[170] Drug registration reforms, including new regulatory circulars such as MoH Circular No. 12/2025/TT-BYT, are intended to speed up approval timelines and are expected to gradually increase the number of orphan medicines available in the domestic market.[171]

In Vietnam, there are an estimated 100 rare diseases affecting approximately 6 million people.[172] Although a formal national rare-disease list has yet to be established, the Ministry of Health has introduced a list of 62 rare and severe diseases eligible for 100% health insurance coverage without referral, which includes several rare conditions such as bone marrow dysplasia and other bone marrow failure syndromes, antiphospholipid and other rare hypercoagulable disorders, infection-associated hemophagocytic syndromes, complex insulin-dependent and non-insulin-dependent diabetes, and inborn errors of metabolism, including aromatic and branched-chain amino acid and fatty-acid metabolism disorders.[173]

At present, only a limited number of internationally used therapies for rare diseases, especially enzyme replacement therapies and metabolic treatments, are registered in Viet Nam and used in clinical practice. Specialised nutrition for inborn errors of metabolism is available through central paediatric hospitals, but distribution is still uneven across regions. Despite these challenges, Viet Nam has made visible progress, and experts note that its treatment landscape, while not yet comprehensive, is shifting from scarcity toward greater structure in availability.

170 Duy, M., & Lieu, D. (2025, September 17). 6 million Vietnamese affected by rare diseases, health ministry launches action plan. *Tuoi Tre News*. Link
171 Tilleke & Gibbins International. (2025, May 27). *Vietnam Issues New Regulations on Drug Registration*. Tilleke. Link
172 Ministry of Health, Vietnam. (2025). Important progress in the management of rare diseases in Vietnam. Link
173 Ministry of Health, Vietnam. (2025). *Ministry of Health announces 62 rare and severe diseases eligible for 100% health insurance coverage without referral*. Link

- Treatment availability for rare diseases across ASEAN remains uneven due to differing regulatory maturity, clinical capacity, affordability and supply systems.
- Singapore and Thailand provide the broadest most equitable access through strong regulatory pathways and national reimbursement systems.
- Malaysia offers consistent clinical protocols for key conditions and is moving toward standardised national supply through its new policy.
- The Philippines and Viet Nam show partial but improving access via compassionate or special-access pathways and increasing registration of orphan drugs.
- Indonesia remains at an early stage, with only limited therapies available through select hospitals or import permits.
- Overall progress is evident, but access still relies heavily on hospital-level capacity rather than coordinated national systems.
- Across ASEAN, approaches to defining rare diseases remain diverse, with Malaysia having established a formal national rare disease list, while other countries are using more targeted or programmatic lists as part of evolving system development.
- Regional collaboration can help close gaps by sharing regulatory experience, aligning treatment eligibility and clinical guidance, and strengthening supply and procurement planning for rare disease therapies.

6.2 Fit-for-purpose Health Technology Assessment

Across Southeast Asia, the use of HTA is growing as governments move towards more efficient, evidence-based allocation of limited health resources while protecting equity. ASEAN countries, therefore, need to adapt HTA methods to be more flexible and inclusive, acknowledging that standard cost-effectiveness thresholds do not fully reflect the personal, social, and ethical values of rare-disease interventions. This section outlines how each country is developing and applying fit-for-purpose HTA models that blend economic evaluation with broader considerations, such as disease severity, personal, ethical, and social values, patient voice, and feasibility, to support more equitable decision-making across the region.

Indonesia

Indonesia has strengthened its HTA system through the Indonesia Health Technology Assessment (InaHTA) Committee under the Ministry of Health, guided by the General Guideline for Health Technology Assessment in Indonesia (2022). The guideline establishes a six-step process – covering topic selection, assessment, appraisal, reporting, decision-making, and post-implementation monitoring – to support evidence-based inclusion, delisting, or price negotiation of health technologies under the JKN. It adopts a multi-domain evaluation framework that integrates clinical, economic, ethical, and organisational considerations, while promoting stakeholder participation from policymakers, clinicians, academics, and industry. This revision marks a major step toward institutionalising transparent, systematic, and evidence-informed priority setting in Indonesia's health system.[174]

The updated guideline introduces adaptive HTA and real-world evidence approaches to address data limitations, particularly for high-cost and rare-disease therapies. Despite these advances, key challenges remain, including limited methodological capacity, insufficient national cost data, and a lack of detailed modelling guidance. As noted in The Lancet Regional Health – Southeast Asia (2023), the guideline provides a strong policy foundation but requires supplementary method guides and tools to enhance practical implementation. Supported by HITAP and International Decision Support Initiative (iDSI), the Ministry of Health is expanding training, developing cost databases, and reviewing processes to align Indonesia's HTA system with international best practices, ensuring that resource allocation decisions are both equitable and context appropriate.[175,176]

Malaysia

The Working Committee on Rare Diseases Technology Assessment, created as a subcommittee under the National Rare Disease Committee, is responsible for evaluating health technologies for rare diseases using the best available evidence on effectiveness, safety, cost implications, and wider health system impact. Technologies are identified at different points in their life cycle, either through horizon scanning for those not yet on the market or through standard HTA processes for new market entries and may later be included in relevant clinical practice guidelines. In 2022, a dedicated framework using Multi-Criteria Decision Analysis (MCDA) was developed to weigh clinical, societal, feasibility, and cost factor in reimbursement decisions for rare disease treatments. After

174 Suryawati, S., et al. (2022). *General Guideline for Health Technology Assessment in Indonesia*. Health Policy Agency Publishing Company. Link

175 Chavarina, K. K., Faradiba, D., & Teerawattananon, Y. (2023). Navigating HTA implementation: a review of Indonesia's revised HTA guideline. *The Lancet Regional Health - Southeast Asia*, *17*, 100280. Link

176 International Decision Support Initiative. (n.d.). *Indonesia*. iDSI. Link

identification by the National Rare Disease Committee, assessments are conducted by the Malaysian Health Technology Assessment Section (MaHTAS), appraised through deliberation and MCDA-assisted voting, and finally endorsed by the HTA and Clinical Practice Guidelines (CPG) Council, chaired by the Director General of Health.

The Philippines

The Philippines has institutionalised HTA through the Universal Health Care Act (RA 11223) and through HTAC, with published process and methodological guides.[177] For rare diseases, this structure is already fit for purpose because it brings together economic evaluation with ethical, social, legal, and system considerations in a deliberative process that can adapt to limited evidence. Topics are nominated by the DOH or PhilHealth nominates topics, and HTAC provides coverage recommendations that combine cost-effectiveness, budget impact, and non-economic criteria such as disease severity and unmet need, which is especially important for orphan therapies in small populations.[178] Although the flexible deliberative process can support decisions more adapted for rare disease situations, there is no consistently applied HTA framework specific for rare disease therapies, such as rarity modifiers, flexible evidentiary standards or separate cost-effectiveness threshold.

Singapore

Singapore's Agency for Care Effectiveness (ACE) is the national body for HTA and clinical guideline development under the Ministry of Health. It evaluates drugs, medical devices, diagnostics, and clinical services to inform subsidy and policy decisions. ACE carries out HTAs through two advisory committees: the Drug Advisory Committee for pharmaceuticals and the Medical Technology Advisory Committee for medical technologies. The process has three stages: topic submission and selection, technical evaluation, and decision-making. Higher-cost technologies with an expected annual budget impact above SGD 2 million receive full evaluations that include systematic reviews and local economic modelling, while lower-cost technologies undergo quicker reviews based on existing evidence and international cost-effectiveness studies. The evaluation system is supported by value-based pricing to ensure public spending is directed toward cost-effective technologies in Singapore's context.[179]

177 Republic of the Philippines. (2019). Republic Act No. 11223 *An Act Instituting Universal Health Care For All Filipinos, Prescribing Reforms in The Health Care System, and Appropriating Funds Therefor. Gazette.* Link

178 Department of Health Philippines. (2020). *Philippine HTA Methods Guide: Methodological standards in evaluation of health technologies in the Philippines.* Health Technology Assessment Unit. Link

179 Agency for Care Effectiveness. (2025, October 15). *Methods and process.* ACE. https://www.ace-hta.gov.sg/resources/process-methods/

ACE also develops ACE Clinical Guidelines (ACGs) through its Evidence to Practice Office (ETPO), which synthesises the best available evidence with local feasibility to provide practical, context-sensitive recommendations for clinicians.[180] These guidelines aim to reduce unnecessary variation in care and prevent costly adverse events, in line with WHO standards for evidence-informed decision-making. Since 2021, ACE has allowed company-led submissions to speed up the evaluation of new drugs nearing regulatory approval, including cancer medicines and, from 2024 onwards, selected non-cancer medicines and cell, tissue, and gene therapy products (CTGTPs). For ultra-rare and high-cost therapies, ACE's detailed HTA process works alongside the RDF,[181] which applies additional criteria, such as disease severity, transformative benefit, and funding sustainability, to support timely and fair access to life-saving treatments.

Thailand

Thailand's HTA ecosystem (HiTAP, NLEM Subcommittee, NHSO) actively uses HTA to support reimbursement decisions. Rare disease therapies are considered under NLEM's E2 category, where inclusion may weigh feasibility, equity, and necessity alongside to cost-effectiveness.[182] Since 2021, several orphan drugs have been added to the NLEM, allowing their procurement and delivery through designated centres. Methodologically, HiTAP continues to update its guidance and commission assessments even when evidence is limited, drawing on international data and modelling. This allows the use of practical approaches, such as limited or temporary inclusion, routine monitoring of real-world use, and gradual expansion of access, which can support the step-by-step integration of rare disease therapies into the NLEM while maintaining budget control.

Viet Nam

Viet Nam has gradually institutionalised Health Technology Assessment (HTA) as a policy instrument to strengthen evidence-based decision-making for health insurance coverage. The Ministry of Health (MOH) first incorporated HTA into the 2013 National Health Strategy. It established a national HTA plan in 2014, followed by regulatory frameworks, such as Decision No. 5315/QD-BYT (2018), which mandate the use of pharmacoeconomic analysis for drugs proposed for health insurance reimbursement.[183] The Health Strategy and

180 Agency for care Effectiveness. (2025). *Ace Clinical Guidances Methods and Processes Manual*. Ministry of Health Singapore. Link

181 Ministry of Health Singapore. (2019, July 2). *Rare Disease Fund to Provide Financial Support to Singaporeans with Rare Diseases* [Press release]. Link

182 Sruamsiri, R., Wagner, A. K., Ross-Degnan, D., Lu, C. Y., Dhippayom, T., Ngorsuraches, S., & Chaiyakunapruk, N. (2016). Expanding access to high-cost medicines through the E2 access programme in Thailand: effects on utilisation, health outcomes and cost using an interrupted time-series analysis. *BMJ open*, *6*(3), e008671. Link

183 Van, M. H., Viet, H. D., & Nu, A. V. (2022). Health technology assessment in Vietnam: current situation and future direction. *Journal of Health and Development Studies*, *06*(04), 9–10. Link

Policy Institute (HSPI) under MOH serves as the national focal point for HTA development, supported by the Department of Health Insurance and the pharmacoeconomic committee established in 2017.

The system is still in an early stage characterised by a limited institutional mandate, insufficiently trained human resources, and a weak data infrastructure. The government is now prioritising the creation of national HTA guidelines, an integrated health data repository, and structured stakeholder participation to enhance transparency and legitimacy in decision-making. As capacity matures, Viet Nam could be positioned to implement fit-for-purpose HTA that integrates social, ethical, and contextual criteria, particularly for high-cost or rare-disease therapies, into reimbursement and pricing decisions to ensure equitable and efficient use of health insurance resources.[184]

- ASEAN state members are increasingly adopting HTA for evidence-based decision making for coverage of treatments. However, there is a need to adapt rigid economic evaluation toward more pragmatic, equity-orientated decision-making for rare diseases.
- Malaysia and the Philippines are starting to incorporate social value judgements into formal appraisal processes.
- Viet Nam and Indonesia are building capacity and adopting adaptive HTA methods and real-world evidence methods.
- Regional collaboration on HTA training, cost databases, and more holistic value assessment and integration of patient voice will be important to support fair, feasible, and meaningful assessments of rare-disease technologies.

184 Lee, H., Nguyen, T. T., Park, S., Hoang, V. M., & Kim, W. (2021). Health Technology Assessment Development in Vietnam: A qualitative study of current progress, barriers, facilitators, and future strategies. *International Journal of Environmental Research and Public Health*, *18*(16), 8846. Link

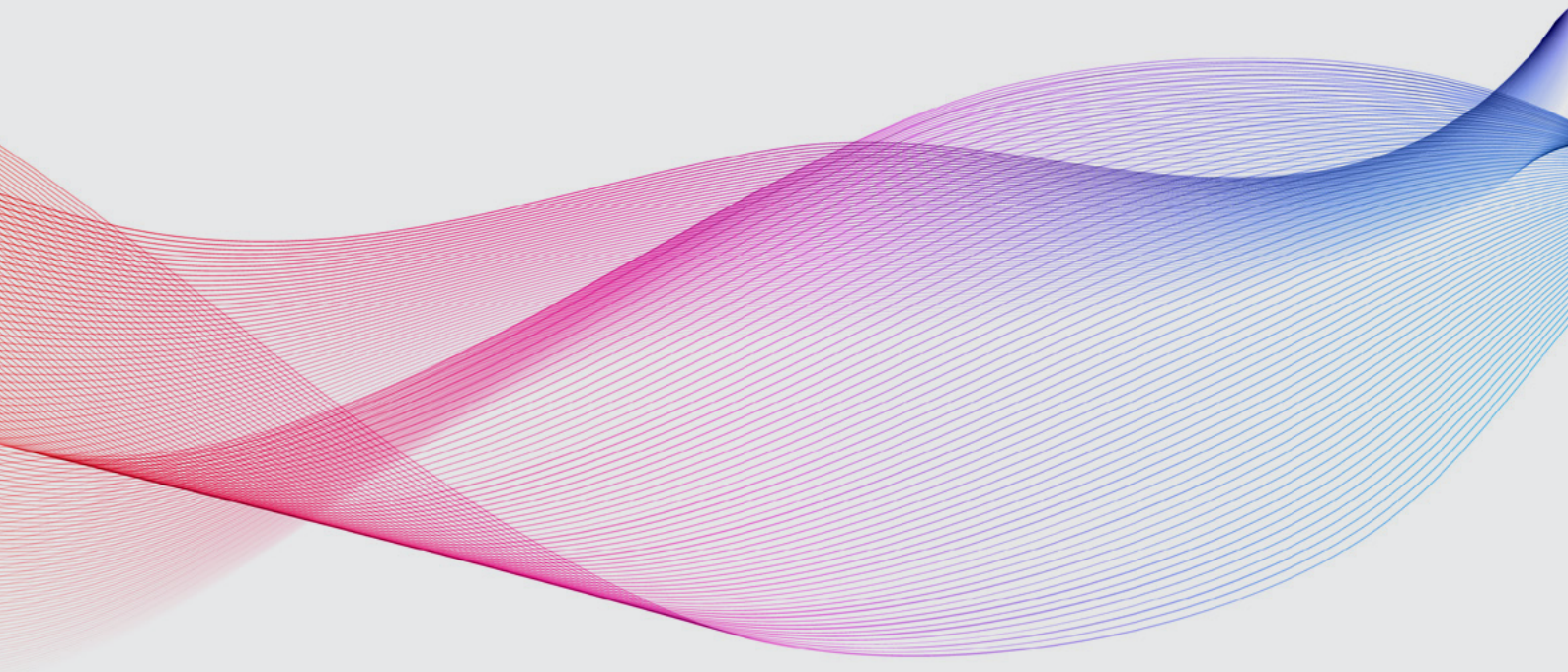
Table 8. Summary of Equitable Pathways to Treatment

Country	Availability of Treatments	Fit-for-Purpose HTA	Regulatory Pathways
Indonesia	Limited availability; access concentrated in a few tertiary hospitals; many therapies depend on import permits, compassionate use, or ad-hoc approvals.	HTA processes still evolving; not yet adapted for rare diseases; limited mechanisms for assessing high-cost, low-volume therapies.	• Accelerated regulatory pathway for rare and serious diseases (100-day review) under BPOM Regulation No. 23/2025 • Special access for importation of unregistered medicines
Malaysia	Provides enzyme replacement therapy for several lysosomal storage disorders; treatment available mainly in tertiary hospitals; structured clinical protocols in place.	HTA exists but not tailored for rare diseases; timelines for orphan drug assessment can be lengthy; no dedicated pathway.	• Standard NPRA regulatory pathways, with orphan medicines increasingly prioritised. • No formal regulatory reliance pathway yet. HTA processes apply, but no dedicated rare-disease HTA framework.
Philippines	Access to several ERTs and orphan medicines in selected tertiary centres; unregistered therapies available through Compassionate Special Permits.	HTA increasing in use but not yet optimised for rare diseases; assessment remains resource-intensive and often slow for specialised therapies.	• CSP mechanism for unregistered orphan medicines. • HTA processes exist nationally, but no rare-disease-specific HTA or fast-track system.
Singapore	Most structured access system; Special Access Route enables use of unregistered medicines; advanced metabolic and genetics services widely available.	HTA is well-established but standard frameworks do not fully accommodate rare diseases; affordability supported through complementary financial schemes.	• SAR allows rapid access to unregistered rare-disease therapies. • Strong regulatory agility through HSA.
Thailand	Strongest public access model; 14 orphan drugs included in the NLEM; nearly half of globally approved orphan drugs are registered; benefits tied to UCS.	HTA integrated into NLEM listing but limited specific provisions for rare diseases; prioritisation linked to UCS cost-effectiveness requirements.	• Structured regulatory and reimbursement pathways through NHSO, NLEM, and national HTA mechanisms. • Rare diseases integrated into benefit package with dedicated assessment and budgeting processes.

Viet Nam	Gradually expanding availability through regulatory reforms; access mainly in specialised paediatric hospitals; limited national supply coordination.	HTA structures developing; no dedicated pathway for rare diseases; pricing and reimbursement decisions remain fragmented.	• MoH Circular No. 12/2025/TT-BYT to accelerate orphan drug registration. • List of 62 rare and severe diseases linked to insurance eligibility.

CHAPTER 7.

Innovative and Sustainable Financing for Rare Disease



CHAPTER 7.

Innovative and Sustainable Financing for Rare Disease

7.1 Universal Health Coverage

Indonesia

Indonesia's UHC system, Jaminan Kesehatan Nasional (JKN), administered by BPJS Kesehatan, seeks to provide comprehensive population coverage through a tiered referral system linking primary care to secondary and tertiary hospitals. While JKN has substantially expanded access to essential health services, the inclusion of rare diseases within UHC remains limited and uneven. Newborn screening for congenital hypothyroidism is mandated, yet national uptake remains very low. Only 4.37% of newborns were screened in 2023, reflecting persistent system and implementation gaps. Expanded screening panels and genetic diagnostics remain largely confined to pilots and tertiary centres. Access to rare disease treatment is not systematically embedded within JKN benefit packages and often depends on hospital capacity, ad hoc financing decisions, compassionate use mechanisms, or external support. As a result, people living with rare diseases continue to face long diagnostic delays, fragmented access pathways, and substantial financial barriers.

Malaysia

Compared with Indonesia, Malaysia has a more established publicly funded service delivery system, providing a stronger foundation for specialised care. Within this system, rare diseases have gained increasing policy visibility, supported by national governance arrangements and expanding specialised services. Public newborn screening currently includes congenital hypothyroidism and G6PD deficiency, and the Institute for Medical Research provides nationwide confirmatory testing for inborn errors of metabolism, following pilot implementation of tandem mass spectrometry. Routine public-sector treatment is available for several rare conditions, including enzyme replacement therapies delivered in tertiary hospitals. However, comprehensive

UHC coverage remains incomplete for advanced genetic testing, newer orphan medicines, and long-term multidisciplinary care, which often rely on special access pathways, hospital-level funding, or charitable support. Malaysia's UHC system therefore offers an important platform for rare disease care, while continuing to face challenges related to sustainability, prioritisation, and equitable nationwide access.

The Philippines

The Philippines reflects a different pathway, where population coverage and early detection are strong, but financial protection across the care continuum remains uneven. The country's pursuit of universal health coverage is anchored in the Universal Health Care Act (Republic Act No. 11223, enacted in 2019) and implemented primarily through PhilHealth, which finances a broad package of preventive and curative services. Rare diseases are legally recognised within the national health framework, and UHC mechanisms have most clearly supported early detection. The Expanded Newborn Screening Programme now covers 29 conditions and has achieved approximately 92% national coverage, a major public health milestone. However, financial protection diminishes beyond screening. Confirmatory diagnostics, specialised genetic testing, and long-term treatments, including enzyme replacement therapies and orphan medicines, are not comprehensively financed and remain concentrated in tertiary hospitals. Consequently, many families continue to face out-of-pocket costs and reliance on special permits or external assistance, illustrating the gap between population coverage and the depth of UHC for rare diseases.

Singapore

Singapore demonstrates how rare diseases are being addressed within a highly structured UHC environment, complemented by targeted financing mechanisms. Singapore's UHC system combines compulsory medical savings, national insurance, tiered public subsidies, and safety nets. Universal newborn screening, introduced nationally in 2006 and progressively expanded, now covers more than 40 disorders and reaches approximately 90% of newborns, or about 40,000 infants each year. Diagnostic services, including confirmatory testing and long-term metabolic monitoring, are embedded within public hospitals and heavily subsidised. Recognising that many orphan medicines exceed standard reimbursement thresholds, Singapore established the Rare Disease Fund, a national public–philanthropic mechanism that provides sustained access to selected high-cost therapies. This model complements UHC by addressing structural financing gaps for ultra-rare and high-cost conditions, while maintaining clinical governance and equity principles.

Thailand

Thailand is another example of a country where rare diseases have been more explicitly incorporated into public financing mechanisms. Thailand's Universal Coverage Scheme, administered by the National Health Security Office, provides comprehensive benefits through an extensive public health network. Newborn screening using tandem mass spectrometry has expanded nationally, with coverage reaching approximately 98% of live births, including outside major urban centres. Under the UCS, 24 rare diseases have been prioritised, supporting diagnostics, confirmatory testing, and long-term management. Selected orphan medicines have been incorporated into the National List of Essential Medicines, enabling public reimbursement. However, financial protection remains largely limited to conditions and products formally listed, leaving patients with other rare diseases to face significant access and affordability barriers.

Viet Nam

Viet Nam illustrates a UHC system with high population coverage, but rare disease integration is still in transition. National prenatal and newborn screening programmes have expanded since 2006, with current screening focused on phenylketonuria, congenital hypothyroidism, and G6PD deficiency. Between 2016 and 2019, national newborn screening coverage increased from 23% to around 40%, although overall coverage remains below 50% and services are concentrated in Hanoi and Ho Chi Minh City. National health insurance partially reimburses screening, but families often still bear the costs of confirmatory and advanced diagnostics. In 2025, major reforms enabled patients with 62 rare and severe diseases to access care at public hospitals with 100% insurance coverage and without referral requirements, marking a significant step toward deeper UHC protection. Nevertheless, advanced genetic services and orphan medicines remain unevenly available and only partially financed, highlighting ongoing challenges in depth of coverage and geographic equity.

Across ASEAN, Universal Health Coverage systems have substantially expanded population access to essential services, but protection for rare diseases remains uneven, with strong progress in newborn screening and selected treatments in countries such as Singapore and Thailand, and more limited, hospital-based or pilot-driven access in Indonesia, the Philippines, and Viet Nam. While several countries have begun integrating rare diseases into public financing and service delivery, significant gaps persist in advanced diagnostics, sustainable financing for orphan medicines, and equitable nationwide access, highlighting the need to strengthen the depth of UHC beyond population coverage.

7.2 Innovative Financing Model

Financing care for rare diseases is a major challenge for health systems in ASEAN, as the costs of diagnosis and lifelong treatment often exceed what insurance schemes and public budgets cover. Sustainable access, therefore, requires new financing models that combine public, private, and philanthropic resources to share financial risk while protecting equity and accountability. Globally, countries use different approaches such as statutory levies, national charity funds, managed entry agreements, and dedicated budget allocations to balance affordability with long-term sustainability. This sub-chapter reviews five such cases, showing how these mechanisms raise new funds, allow flexible reimbursement, and connect social insurance with research and innovation. Together, they offer practical lessons on how ASEAN can build stronger, more resilient financing systems for rare-disease treatment.

National Charity Fund with Government Co-match – Singapore

Singapore's RDF is one of the most structured public-private models for financing lifelong, high-cost treatments for rare diseases. Established in 2019, it is a registered charity that combines philanthropic donations with a 3:1 government matching contribution to subsidise enzyme replacement therapies and other ultra-orphan medicines for eligible Singaporean patients. Treatment decisions are made by a multidisciplinary expert panel that reviews medical need, disease severity, and evidence of effectiveness. The fund focuses on therapies that work but are too expensive for most families, serving as a last-resort option when other financial support is insufficient. As of 31 July 2022, the RDF had raised USD 106 million (S$137.9 million), with long-term sustainability supported by endowment investment returns and strong governance under the Ministry of Health.[185] This co-funding model has enabled patient access to several high-cost rare disease treatments while maintaining fiscal prudence and public accountability.

Public Insurance Reimbursement for Orphan Drugs – Thailand

Thailand provides rare disease treatment through the UCS by using the NLEM, especially category E2, which includes high-cost and orphan medicines. Since 2021, the National Health Security Office has added four orphan drugs to the list, followed by ten more in 2022, ensuring free access under all three public health insurance schemes once a medicine is listed. Inclusion relies on health technology assessments by HITAP and public consultations to maintain transparency. This model builds equity into the main health system rather than creating separate programmes and uses centralised procurement to secure

185 Ministry of Health Singapore. (2022, October 3). *Donations Made To Rare Disease Fund and Alternative Fund-raising Arrangements with Matching Donation Support For More Diseases*. Ministry of Health. Link

lower prices and broader access. The NHSO's regular updates show strong political commitment to expanding rare disease coverage using HTA principles.

Dedicated Budget Line within Universal Health Insurance – Taiwan

Taiwan's National Health Insurance (NHI) provides one of the most comprehensive frameworks for financing rare disease care under the Rare Disease Control and Orphan Drug Act (2000).[186] Patients diagnosed with one of 225 recognised rare diseases receive a catastrophic illness card, exempting them from co-payments for covered diagnostics, medicines, and long-term care. The National Health Insurance Administration (NHIA) earmarks a dedicated annual budget line, which increased from USD 232 million (NT$7.16 billion) in 2020 to USD 285 million (NT$8.78 billion) in 2022,[187] alongside a steady rise in registered patients. The Act mandates cross-ministerial coordination to support research, newborn screening, and drug importation. By combining broad insurance coverage, legal safeguards, and transparent reporting, Taiwan has created one of Asia's most sustainable models for fair access to orphan medicines. It does this by linking social insurance, research, and patient protection within a single legislative framework.

Managed Entry Agreements – South Korea

South Korea pioneered risk-sharing agreements (RSAs) in 2013 to manage reimbursement for high-cost cancer and orphan drugs within its National Health Insurance framework. RSAs allow flexible reimbursement conditions, such as expenditure caps, outcome-based refunds, and price-volume agreements, enabling earlier patient access while controlling financial risk.[188] Evaluations show that RSAs have expanded access to high-cost therapies that were previously unaffordable, sharply reducing patients' out-of-pocket spending and improving equity. The system is managed by the Health Insurance Review and Assessment Service, which reviews real-world evidence during each contract period before renewal. From 2013 to 2022, more than 60 medicines were covered through RSA mechanisms.[189] This structured approach shows that flexible reimbursement can expand access to new, high-cost treatments while still keeping overall spending under control.

186 Hsiang, N., Huang, W., Gau, C., Tsai, T., & Chang, L. (2021). The impact of The Rare Disease and Orphan Drug Act in Taiwan. *Journal of Food and Drug Analysis*, *29*(4), 717–725. Link

187 National Health Insurance Administration. (2022, September 16). *The NHIA stands with rare disease patients, sparing no effort in their care*. Ministry of Health and Welfare. Link

188 Lee, B., Bae, E., Bae, S., Choi, H., Son, K., Lee, Y., Jang, S., & Lee, T. (2021). How can we improve patients' access to new drugs under uncertainties? : South Korea's experience with risk sharing arrangements. *BMC Health Services Research*, *21*(1). Link

189 International Society for Pharmacoeconomics and Outcomes Research. (2023, December). *Have risk sharing agreements in South Korea achieved the intended policy objectives?* ISPOR. Link

Statutory Industry Levy for Rare Disease Access and Research – Italy

Italy's AIFA "5% Fund", created under Law 326/2003, is a legally required levy that collects 5% of all pharmaceutical promotional spending to support early access to orphan drugs and independent clinical research. The Italian Medicines Agency manages the Fund, which finances compassionate-use access for treatments not yet reimbursed and provides grants for investigator-led rare-disease studies. Law 175/2021 expanded the mechanism by adding another 2% dedicated specifically to rare-disease research and nonprofit clinical trials.[190] The mechanism's revenue depends on how much companies spend on marketing, creating a steady funding stream while maintaining public oversight. Since it began, the 5% Fund has supported hundreds of independent research projects and many early-access programmes, making it one of the longest-running public–private models that link access, innovation, and transparency in rare-disease policy. Between 2005 and 2018, AIFA issued nine calls for independent research grants and funded 282 clinical studies, including 111 studies on rare diseases or rare tumours, with a total investment of USD 50.4 million (€43.46 million).[191]

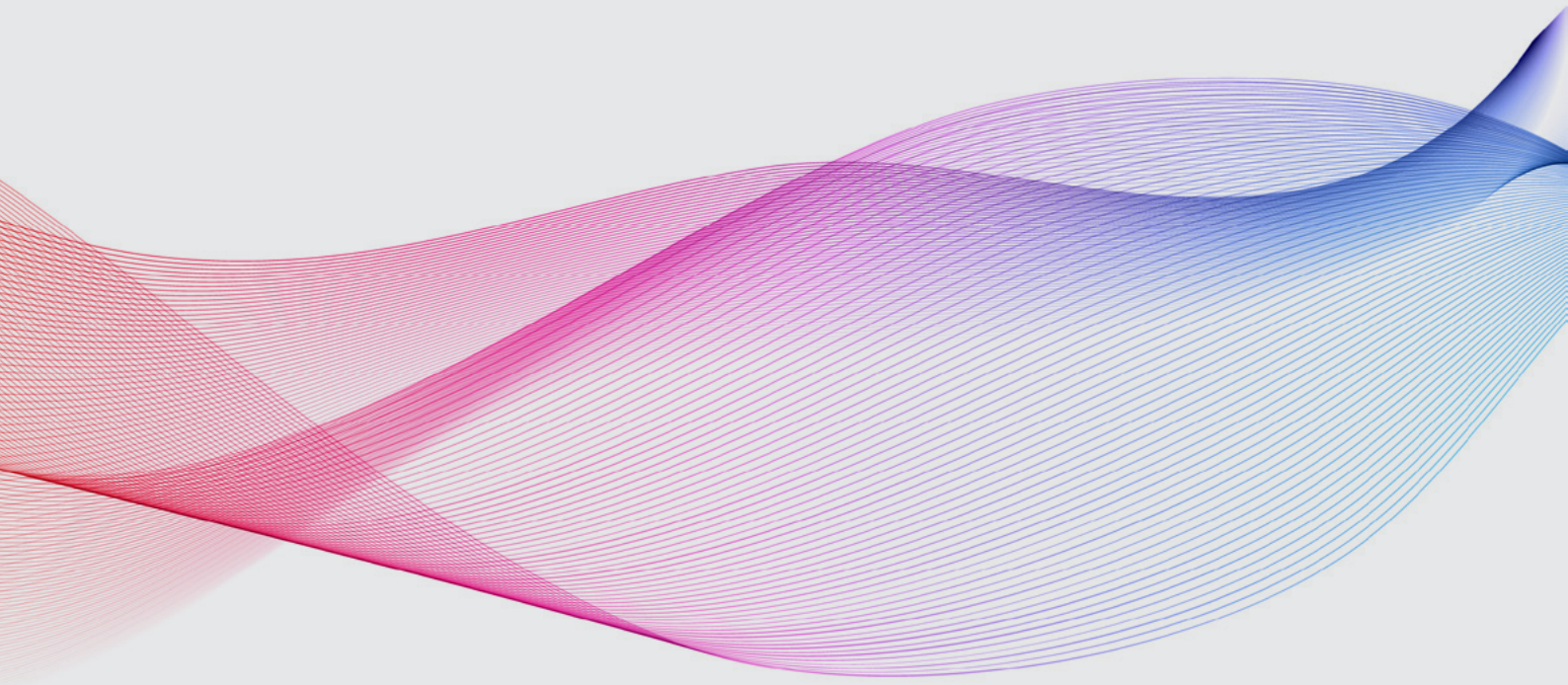
- Sustainable financing for rare diseases is achievable when equity, transparency and shared responsibility guide the design of funding models.
- Examples from Singapore, Thailand, Taiwan, South Korea and Italy show how coordinated mechanisms can provide predictable support for people with lifelong conditions.
- Adapting such approaches in ASEAN will require phased experimentation, clear governance and partnerships that align public funding, private investment and community contributions.
- Innovative financing is ultimately about creating fair and durable systems that protect patients from financial hardship while ensuring access to effective treatment.

190 Agenzia Italiana Del Farmaco. (2025, April 11). *AIFA Board approves the call for independent research on rare diseases for 2025*. AIFA. Link

191 Marconi, P., Zappalà, C., Lofaro, A., Maiolino, S., Mastellone, C., & Belfiglio, M. (2022). [The independent research on rare diseases financed by the Italian Medicines Agency.]. *PubMed*, *113*(7), 451–459. Link

CHAPTER 8.

Regional Priority Areas



CHAPTER 8.

Regional Priority Areas

The Southeast Asia Rare Diseases Policy Forum held in Putrajaya on 7-8 November 2025 reinforced a shared regional understanding of the need to address persistent gaps in rare disease diagnosis, care, and financing through closer cooperation among ASEAN Member States.

Drawing on the priorities identified by governments, patients, clinicians, and other stakeholders, this chapter consolidates key regional priorities and presents practical, forward-looking recommendations to support both national system strengthening and regional collaboration. These priorities are intended to inform a more coherent ASEAN approach to rare diseases, enhancing early detection, expanding equitable access to treatment, and fostering sustainable, people-centred responses across the region. In doing so, they also provide a substantive foundation to inform discussions on a potential ASEAN Rare Disease Declaration.

1. Integrate Rare Diseases into ASEAN's Health Agenda and Build Long-term Regional Solidarity

ASEAN should take concrete steps to embed rare diseases within its existing health cooperation mechanisms to sustain long-term dialogue and action:

- Embed rare diseases under the ASEAN Health Clusters, including noncommunicable diseases, universal health coverage (UHC), and pharmaceutical development, to ensure sustained dialogue within the Region.
- Establish an ASEAN coordination mechanism (e.g., ASEAN Rare Disease Platform) or working group and leverage existing dialogue platforms (e.g., ASEAN ministerial meetings and health cluster agendas) to promote policy alignment, regulatory harmonisation, data collaboration, knowledge and technical exchange, and joint advocacy.
- Develop joint capacity-building and collaborative research networks across ASEAN countries and dialogue partners (e.g., Japan, Australia) in policy, genomics, epidemiology, health economics, and clinical trials.

- Encourage each Member State to develop or update national rare disease frameworks with measurable targets and monitor progress through shared indicators aligned with the ASEAN Health Development Framework.
- Develop and harmonise regional registries, data standards, governance frameworks, and coding systems to enable ethical and interoperable regional data sharing, and establish cross-border referral mechanisms where domestic capacity is limited.

2. Strengthen Early Detection and Diagnosis

Countries should prioritise early identification of rare diseases through strengthened screening systems, genomic capacity, and multi-stakeholder collaboration:

- Expand newborn screening (NBS) programmes and integrate genomic testing into national health systems.
- Develop national registries to understand the disease burden and guide effective interventions. Build diagnostic and genomic workforce capacity, including laboratory, pathology, and bioinformatics specialty training.
- Promote public-private partnerships to accelerate innovation in diagnostics and therapeutics.
- Explore research and emerging technologies for presymptomatic and carrier screening to support early detection and prevention of rare diseases.

3. Improve Equitable Pathways to Treatment

To enhance access to timely, high-quality treatment across the region, ASEAN Member States should:

- Expand telemedicine to improve treatment access for underserved populations across member states, while leveraging AI and data analytics for diagnosis, disease mapping, and health system planning.
- Establish centres or hubs of excellence to support timely diagnostic, counselling and clinical care within the country and across the ASEAN regions.
- Ensure a patient-centred approach in the rare disease care pathway by enacting non-discrimination and data protection policies that safeguard patients' rights, institutionalising patient participation in national and regional governance structures, and empowering patient organisations to engage in advocacy, research, and peer support.

- Incorporate quality-of-life and social inclusion indicators in monitoring frameworks. This also includes integrating the voices of families and caregivers throughout the patient journey from diagnosis to treatment.
- Raise public awareness to reduce stigma and promote early health-seeking behaviour by harnessing patients' voices.

4. Explore Innovative and Sustainable Financing for Rare Disease Management

To reduce financial burden and enable long-term sustainability of rare disease programmes, ASEAN should:

- Integrate rare diseases into UHC benefit packages to reduce out-of-pocket burden and develop transparent prioritisation frameworks to guide coverage and ensure long-term financial sustainability.
- Promote the adoption of innovative and context-appropriate financing mechanisms across ASEAN Member States, such as risk-sharing or Managed Entry Agreement (MEA) models for high-cost therapies, a copayment model with the private sector, and the establishment of dedicated rare disease funds through blended public-private-philanthropic approaches, with alternative funding sources such as faith-based and community-based sources.
- Explore a pooled procurement mechanism to expand access to therapy across the region.

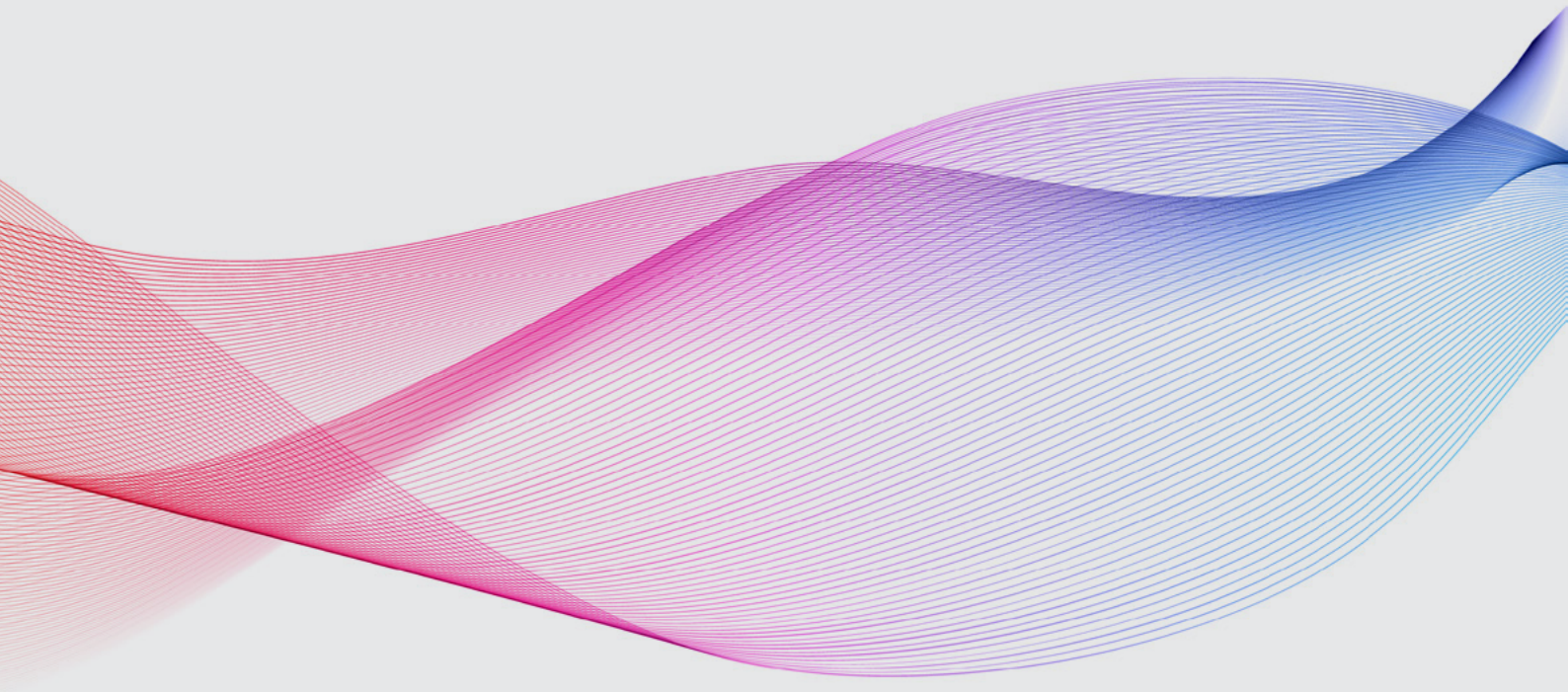
5. Strengthen National Governance for Rare Diseases Across Member States

Countries should reinforce institutional governance to improve coordination, accountability, and multi-sectoral support for rare-disease communities:

- Ensure that dedicated units within health ministries are responsible for coordinating rare disease policies and financing.
- Integrate rare disease programmes into multi-sectoral initiatives by involving education, social protection, and research agencies.
- Include participation of patients, families, and caregivers in policy development and implementation to ensure the quality of life of patients and caregivers.

CHAPTER 9.

Conclusion



CHAPTER 9.
Conclusion

ASEAN rare-disease systems are fragmented but show examples of rapid progress. Embedding rare disease priorities within existing health cooperation mechanisms and advancing an ASEAN Rare Disease Declaration and Action Plan can institutionalise regional collaboration and accelerate national reforms. The two-stream framing and ASEAN-specific roadmap provide a pragmatic foundation for action that is both feasible and aligned with global commitments.

Furthermore, this report's findings show that rare diseases remain a significant yet addressable challenge in Southeast Asia. Although the six ASEAN Member States reviewed in this study have distinct health system structures and policy environments, they face common gaps. These include long delays in diagnosis, limited genomic capacity, the absence of comprehensive national registries, insufficient referral coordination, lengthy access pathways and major financial barriers to accessing high-cost therapies. These gaps contribute to inequities for PLWRD and their families, who often experience fragmented care pathways and substantial socioeconomic burden.

At the same time, momentum for action is clearly growing. Several countries have taken initial steps that can serve as foundations for regional progress. The Philippines has established a legal framework that defines rare-disease care as a state responsibility. Malaysia's new NPRD creates a structured platform for patient-centred services, improved governance, and expanded diagnostic and treatment capacity. Thailand has integrated rare diseases into its universal health coverage programme and continues to scale genomic capacity through national initiatives. Singapore has demonstrated how public and community contributions can expand access to essential therapies. Viet Nam's expanded insurance coverage for more than 62 rare and severe conditions marks a significant advancement toward equitable access. Indonesia's national genome initiatives and its regulatory pathway for orphan drugs illustrate growing recognition of the need for rare-disease reform.

Despite these advances, regional progress will require stronger cooperation. Countries can benefit from shared coding and classification standards,

coordinated efforts to strengthen reference laboratories, and pilot activities to support mutual reliance in regulatory processes for orphan drugs. Collaboration on cost information, procurement experience, and price transparency can help reduce financial barriers. Patient organisations can be key contributors to address some of these challenges, through more inclusive consultation mechanisms and regional engagement platforms. A simple cooperation dashboard can promote accountability by tracking progress across ASEAN in a practical and low-burden way.

The Southeast Asia Rare Diseases Policy Forum in November 2025 demonstrated strong willingness among governments, patient groups, clinicians, researchers, industry and partners to collaborate on solutions. As ASEAN develops its Post-2025 Vision and future health cooperation frameworks, rare diseases can be integrated into existing priorities that focus on universal health coverage, health system strengthening, innovation, and equity. By aligning national actions with regional commitments and global guidance, ASEAN can make meaningful progress toward earlier diagnosis, more equitable access to care, and improved quality of life for people living with rare diseases across the region.

As all UN Member States work to implement the WHA Resolution at the national level, the WHO will also be developing the GAPRD, which will provide a tangible framework for action at all levels. To do this successfully, the WHO will need to consult relevant stakeholders for input. The outcomes of the Southeast Asia Rare Diseases Policy Forum inform the development of the GAPRD by coordinating regional perspectives and input, alongside the Western Pacific Regional Task Force, and the Southeast Asia Regional Task Force established by RDI, grounded in the realities of ASEAN and other low- and middle-income settings. The approaches outlined in this report reflect health systems characterised by constrained resources, evolving diagnostic capacity, and the need to balance population-level investments with fit-for-purpose access to high-cost therapies. By articulating pragmatic pathways shaped by these contexts, the report provides perspectives that can complement global deliberations and help ensure that the forthcoming Global Action Plan is responsive to diverse system realities beyond high-income settings.

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List of Abbreviations

AHMM : ASEAN Health Ministers Meeting
AMR : Antimicrobial Resistance
APARDO : Asia Pacific Alliance of Rare Disease Organisations
APEC : Asia-Pacific Economic Cooperation
ASCC : ASEAN Socio-Cultural Community
ASEAN : Association of Southeast Asian Nations
BGSi : Biomedical and Genome Science Initiative
BRIN : Badan Riset dan Inovasi Nasional / National Research and Innovation Agency
CAH : Congenital Adrenal Hyperplasia
CH : Congenital Hypothyroidism
COMP : Committee for Orphan Medicinal Products
CSP : Compassionate Special Permit
DOST : Department of Science and Technology
ENBS : Expanded Newborn Screening (Philippines)
EMA : European Medicines Agency
ERT : Enzyme Replacement Therapy
ERNs : European Reference Networks
GAPRD : Global Action Plan on Rare Diseases
G6PD : Glucose-6-Phosphate Dehydrogenase
GISAID : Global Initiative on Sharing All Influenza Data
HKL : Hospital Kuala Lumpur
HTA : Health Technology Assessment
ICD : International Classification of Diseases
IMR : Institute for Medical Research
IRDiRC : International Rare Diseases Research Consortium
JACG : Joint Assessment Coordinating Group
JKN : *Jaminan Kesehatan Nasional* (Indonesia's National Health Insurance)
KKH : KK Women's and Children's Hospital (Singapore)
MGVI : Malaysia Genome & Vaccine Institute
MoH : Ministry of Health
MOSTI : Ministry of Science, Technology and Innovation

NCDs : Noncommunicable Diseases
NBS / NBSCC : Newborn Screening / Newborn Screening Continuity Clinics
NBSRC : Newborn Screening Reference Centre
NEHR : National Electronic Health Record
NGS : Next-Generation Sequencing
NHI : National Health Insurance
NHSO : National Health Security Office
NLEM : National List of Essential Medicines
NIH : National Institutes of Health Malaysia
NPRA : National Pharmaceutical Services Programme
NPRD : Malaysia's National Policy for Rare Diseases (2025)
OOP : Out-of-pocket
PGC : Philippine Genome Center
PLWRD : People Living with Rare Diseases
PPWG : Pharmaceutical Product Working Group (ASEAN)
RDF : Rare Disease Fund
RDI : Rare Diseases International
RSCM : Cipto Mangunkusumo National Central Hospital (Indonesia)
SAS : Special Access Scheme
SAR : Special Access Route
SDGMC : SingHealth Duke-NUS Genomic Medicine Centre
SDGs : Sustainable Development Goals
SG10K/SG100K : Singapore 10,000 Genomes / 100,000 Genomes Initiatives
SIKNAS/SIKN : *Sistem Informasi Kesehatan Nasional* (National Health Information System)
SMA : Spinal Muscular Atrophy
T&CM : Traditional and Complementary Medicine
T-RUN : Thailand Rare and Undiagnosed Disease Network
UCS : Universal Coverage Scheme
UHC : Universal Health Coverage
UNGA : United Nations General Assembly
WHO : World Health Organization
WHA : World Health Assembly
WGS : Whole Genome Sequencing

ANNEXURE 1.

Global WHO Policy Frameworks Relevant to Rare Diseases

- **Universal Health Coverage (UHC)**[192]

  WHO defines UHC as ensuring that all people have access to the full range of quality health services they need, across health promotion, prevention, treatment, rehabilitation, and palliative care, without experiencing financial hardship. This framing is directly relevant to rare diseases, as it establishes the normative basis for including diagnostics, long-term care, and high-cost services within essential benefit packages while addressing catastrophic out-of-pocket expenditure.

- **Integrated People-Centred Health Services**[193]

  The WHO people-centred care framework calls for health systems to be organised around the needs of individuals and communities rather than diseases or institutions, with emphasis on continuity, coordination, and participation. This approach closely aligns with the needs of patients with rare diseases, who often navigate fragmented pathways across multiple providers over the life course.

- **WHO Framework on Noncommunicable Diseases (NCDs)**[194]

  WHO characterises NCDs as chronic conditions of long duration and slow progression that require sustained management rather than episodic care. Although rare diseases are distinct, many share similar characteristics of lifelong care needs and cumulative burden, making NCD policy principles relevant for health-system planning and service delivery models.

- **Integrated Management of NCDs and WHO PEN**[195]

  The WHO Package of Essential Noncommunicable Disease Interventions (PEN) focuses on strengthening primary health care in low-resource settings through integrated, cost-effective service delivery. Its emphasis on early detection, referral, and continuity of care offers a relevant model for integrating rare disease pathways within primary care and referral systems in low- and middle-income contexts.

192 World Health Organization. (2019, July 16). *Universal health coverage*. Link
193 World Health Organization. (2020, December 8). *Integrated people-centred care - GLOBAL*. Link
194 World Health Organization. (2025, September 25). *Noncommunicable diseases*. Link
195 World Health Organization. (n.d.). *Integrated management of NCDs*. Link

- **WHO Genomics Programme**[196]

  WHO positions genomics as a public health tool that supports disease prevention, diagnosis, and management, rather than as a narrowly specialised or tertiary intervention. In the context of rare diseases, genomics plays a critical role in reducing diagnostic delays and enabling more precise clinical decision-making when embedded within health systems.

- **WHO Science Council Report on Accelerating Access to Genomics**[197]

  The WHO Science Council identifies genomics as essential for improving population health globally and underscores that delayed access in low- and middle-income countries is neither ethically nor scientifically justifiable. The report highlights promotion, implementation, collaboration, and ethical governance as core pillars for ensuring that genomic advances translate into equitable health gains, including for rare diseases.

- **WHO Guidance on Human Genome Data Collection, Access, Use and Sharing**[198]

  WHO sets out globally applicable principles for the ethical, equitable, and responsible governance of human genome data, grounded in human rights, social justice, and public trust. These principles are particularly important for rare diseases, where data sharing, registries, and cross-border collaboration are essential but must be balanced with safeguards for equity, consent, and accountability.

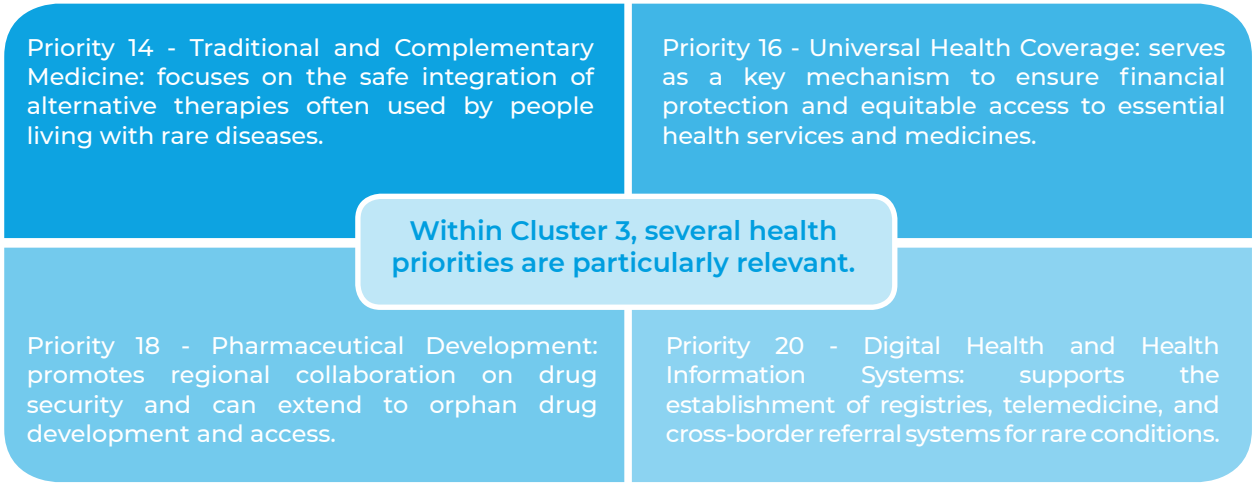
196 World Health Organization. (2023, September 11). *Genomics - Global*. Link
197 World Health Organization. (2022, July 12). *Accelerating access to genomics for global health: promotion, implementation, collaboration, and ethical, legal, and social issues: a report of the WHO Science Council*. Link
198 World Health Organization. (2024, November 20). *Guidance for human genome data collection, access, use and sharing*. Link

ANNEXURE 2.
ASEAN Commitments

4.2.1 ASEAN Post-2015 Health Development Agenda 2021-2025

The APHDA provides ASEAN's operational framework for health cooperation. **Cluster 3: Strengthening Health Systems and Access to Care** offers the clearest entry point for rare diseases, given its focus on UHC, affordable and quality care, essential medical supplies (including traditional and complementary medicines), innovation, digital health, and services for vulnerable population groups. These priorities naturally align with the principles of **WHA Resolution 78.11**, which calls for integrating rare diseases into UHC and strengthening diagnostics and equitable access to care.[199]

Cluster 1: Promoting Healthy Lifestyles, also provides an important complementary entry point for rare diseases. While traditionally focused on prevention and health promotion, its emphasis on community engagement, public awareness, early health-seeking behaviour, and addressing social determinants of health aligns closely with the needs of rare-disease communities. Strengthening communication, reducing stigma, and empowering families to recognise early signs of rare conditions can support earlier diagnosis and improve inclusion within broader public-health initiatives. Integrating these elements into rare-disease strategies can help ensure that community-level interventions reinforce system-level reforms under Cluster 3.



199 ASEAN Secretariat. (2023). *ASEAN Post-2015 Health Development Agenda (APHDA) (2021-2025)*. ASEAN Secretariat. Link

4.2.2 ASEAN Leaders' Declaration on One Health Initiative and ASEAN Declaration of Commitment on Drug Security and Self-Reliance

The ASEAN Charter formally enshrined ASEAN centrality as a guiding principle of the regional organisation. Under Article 1 on "Purposes," Point 15 states that ASEAN seeks "to maintain the centrality and proactive role of ASEAN as the primary driving force in its relations and cooperation with its external partners in a regional architecture that is open, transparent and inclusive." In parallel, Article 41 on "Conduct of External Relations," Point 3 affirms that "ASEAN shall be the primary driving force in regional arrangements that it initiates and maintains its centrality in regional cooperation and community building."

The Charter subsequently became one of the foundational blocks for the 42nd ASEAN Summit in 2023, where leaders adopted the ASEAN Leaders' Declaration on the One Health Initiative, followed by the 2024 launch of the ASEAN One Health Network (AOHN) and the One Health Joint Plan of Action (OH JPA). These instruments strengthen multisector coordination, surveillance, preparedness, and cross-border cooperation. In 2025, ASEAN further adopted the ASEAN Declaration of Commitment on Drug Security and Self-Reliance (ADSSR), extending cooperation on the medicine life cycle, from production and regulation to procurement and distribution; covering essential, high-cost, and non-commercially viable medicines, including those needed for rare diseases. Together, these frameworks illustrate ASEAN's growing commitment to shared responsibility, joint investment, and resilient health systems. By embedding rare diseases into future declarations and cooperation mechanisms, ASEAN can ensure stronger visibility, national follow-up, and accountability across Member [200][201][202]

200 ASEAN Secretariat. (2023, May 10). *ASEAN Leaders' Declaration on One Health Initiative*. ASEAN. Link
201
202 Public Relations of BKPK. (2024, June 21). *Launch of the ASEAN One Health Network and the ASEAN One Health Joint Plan of Action*. KemenkesBKPK. Link

4.2.3 ASEAN Pharmaceutical Product Working Group (PPWG) and Joint Assessment Coordinating Group (JACG)

The Pharmaceutical Product Working Group (PPWG) was established following the recommendation of the 13th ASEAN Consultative Committee for Standards & Quality (ACCSQ) Meeting that took place from 18 to 19 March 1999 in Manila and endorsed by SEOM (Senior Economic Officials Meeting) in January 2000 to advance economic integration for pharmaceutical products without compromising safety, efficacy and quality.[203] Since its formation, the PPWG has progressed key harmonisation initiatives, including the ASEAN Common Technical Dossier (ACTD) and ASEAN Common Technical Requirements (ACTR) concluded in 2008, the ASEAN Sectoral Mutual Recognition Arrangement for GMP Inspection signed in April 2009, and the ASEAN Sectoral MRA on Bioequivalence Study Reports concluded in 2016 and signed in 2017. These measures form the regulatory foundation for the implementation of the ASEAN Pharmaceutical Regulatory Policy (APRP), which was adopted by ASEAN Economic Ministers and ASEAN Health Ministers in 2022 to advance harmonised standards, strengthen cooperation among National Regulatory Authorities and ensure timely access to high-quality pharmaceutical products.[138] The APRP also provides the basis for the ASEAN Pharmaceutical Regulatory Framework (APRF), which was endorsed in 2022 and adopted intersessionally by AHMM on 31 March 2023 and AEM on 5 April 2023.[204]

Within this broader regulatory ecosystem, the Joint Assessment Coordinating Group (JACG) functions under the guidance of the PPWG to facilitate cooperation among National Regulatory Authorities through joint assessment of marketing authorisation applications.[205] The joint assessment procedure is a coordinated process in which the same application is simultaneously submitted to all participating ASEAN NRAs, with assessments conducted collectively and consolidated into a joint assessment report that is then used for national decision-making. Participation is voluntary and requires a minimum of three NRAs. Priority product categories for the ASEAN Joint Assessment include treatments for HIV, TB, malaria, hepatitis B and C, cancer, maternal and reproductive health, and rare diseases, with "products for the treatment of rare diseases (orphan drugs)" explicitly included in the revised 2022 priority list.[206]

- Ultimately, the PPWG and JACG provide ASEAN with a strong foundation to improve access to rare disease treatments.

203 ASEAN Secretariat. (n.d.). *Frequently Asked Questions (FAQ) on the ASEAN Joint Assessment (JA) Procedure*. Link
204 ASEAN Secretariat. (2022). *ASEAN Pharmaceutical Regulatory Policy*. Link
205 ASEAN Secretariat. (n.d.). *Frequently Asked Questions (FAQ) on the ASEAN Joint Assessment (JA) Procedure*. Link
206 ASEAN Secretariat. (n.d.). *List of Priority Product Types/Categories for ASEAN Joint Assessment Procedure: Revised on 24 February 2022*. Link

- The PPWG develops common regulatory standards and harmonised requirements that help countries evaluate medicines consistently and efficiently, which is important for rare disease therapies that are often complex and limited in supply.
- The JACG builds on this by enabling ASEAN countries to jointly assess the same rare disease medicine through a single coordinated review. This reduces duplication, speeds up decision-making and allows countries to rely on shared expertise.
- Together, these mechanisms support faster, more aligned and more reliable access to orphan drugs across the region.

4.2.4 Best Practices and Innovation: Lessons for Rare-Disease Integration

The ASEAN publication *"Strengthening Health Systems and Access to Care (2019)"* showcases a wide range of national innovations that demonstrate how countries in the region are improving health service delivery through efficiency, collaboration, and innovation.[207] While these initiatives were not created specifically for rare diseases, they provide strong evidence of how existing systems can be leveraged to support care for people living with rare conditions. Each of these experiences addresses issues central to rare diseases, such as delayed diagnosis, limited specialist access, and uneven health coverage. By studying these examples, ASEAN can learn how to embed rare-disease responses within ongoing health system reforms rather than treating them as separate, high-cost programmes.

Table 9. Overview of Health System Good Practices in ASEAN

Country & Innovation	Why This Matters for Rare Diseases
Indonesia's Emergency Medical Service 119	Supports faster referrals and coordinated action, which can reduce delays in reaching specialist centres.
Malaysia's Cluster Hospital Initiative	Expands access to specialist expertise and improves diagnostic accuracy, which is essential for molecular and genetic conditions.
The Philippines' Distributed Radiology Model	Enables remote facilities to access expert imaging, reducing diagnostic delays for complex conditions.

207 ASEAN Secretariat. (2019). *Strengthening Health Systems and Access to Care: Best Practices in ASEAN*. Link

Singapore's Team-Based Primary Care	Provides coordinated, long-term management for complex, multi-system conditions common in rare diseases.
Thailand's Supply Chain management	Ensures reliable distribution of high-cost or limited-quantity medicines, including orphan treatments.
Viet Nam's Service Coverage Expansion	Strengthens financial protection and local access to essential diagnostics, reducing the burden on affected families.

Together, these examples show that ASEAN countries already possess the building blocks for integrating rare-diseases. The region's focus on health system strengthening, universal coverage, and innovation aligns naturally with the needs of people living with rare diseases. Rather than creating separate mechanisms, ASEAN can embed rare-disease care within existing health reforms by applying the principles demonstrated in these best practices: collaboration, inclusivity, and shared learning. Doing so will advance not only the rare-disease agenda but also ASEAN's broader commitment to equity, resilience, and the vision of a truly people-centred community.

4.2.5 Moving Toward an ASEAN Framework for Rare Diseases

Taken together, the Hanoi Declaration, the APHDA 2021 to 2025, the Joint Report on Primary Health Care and T&CM, the ASEAN Leaders' Declaration on the One Health Initiative, and the ASEAN Declaration of Commitment on Drug Security and Self-Reliance form a coherent regional framework that aligns closely with global rare-disease commitments. These instruments share principles of people-centred development, innovation, equity and shared responsibility, which also underpin United Nations Resolution 76/132 (2021) and World Health Assembly Resolution 78.11 (2025). The One Health Initiative and the subsequent establishment of the ASEAN One Health Network and Joint Plan of Action demonstrate how ASEAN can translate high-level commitments into coordinated, multisectoral mechanisms that strengthen surveillance, preparedness and cross-sector data-sharing, lessons that are relevant for early detection and coordinated care for rare diseases. Similarly, the Drug Security and Self-Reliance Declaration, adopted in 2025, reinforces equitable access to essential and high-cost medicines by strengthening production, regulation, procurement and distribution processes, offering a clear pathway to improve regional access to orphan and non-commercially viable medicines needed for rare-disease care.

As ASEAN develops its Post-2025 Vision, there is a clear opportunity to mainstream rare diseases within existing regional priorities rather than treating them as a niche issue. This can be achieved by:

1. Embedding rare-disease strategies under Cluster 3 or Cluster 1 and its future equivalent within the next APHDA cycle.
2. Leveraging digital health cooperation for regional registries, teleconsultation, and data-driven decision-making.
3. Integrating rare-disease indicators into Primary Health Care performance monitoring.
4. Promoting cross-border collaboration in pharmaceutical security and orphan drug access.

Furthermore, ASEAN already hosts specialised centres such as the ASEAN Centre for Active Ageing and Innovation (ACAI), the ASEAN Centre for Biodiversity (ACB), the ASEAN Centre for Public Health Emergencies and Emerging Diseases (ACPHEED), and the ASEAN Coordinating Centre for Humanitarian Assistance (AHA Centre) which demonstrate the effectiveness of institutionalised regional mechanisms. Building on this model, ASEAN could consider establishing an ASEAN Centre for Rare Diseases (ACRD) under Health Cluster 3. The ACRD could coordinate regional registries, facilitate genomic and clinical collaboration, and potentially harmonise diagnostic and regulatory standards. Such a centre would transform fragmented national initiatives into a shared ASEAN platform for equity, innovation, and sustainability, aligning directly with the WHA Resolution 78.11 and the forthcoming Global Action Plan on Rare Diseases.

ANNEXURE 3.

ASEAN Member State Highlights and Opportunities for Cross-Learning

4.3.1 Indonesia

Indonesia has not yet enacted a specific law or national strategy dedicated to rare diseases, and relevant provisions are embedded within broader health and social-protection frameworks. Several regulations provide an indirect policy foundation, including Minister of Health Regulation No. 71 of 2015 on the Control of Non-Communicable Diseases, which may include certain rare disorders, Coordinating Ministry for Human Development and Cultural Affairs Regulation No. 7 of 2022 on inter-ministerial coordination of health programmes, and Government Regulation No. 28 of 2024 implementing Health Law No. 17 of 2023, which guarantees equitable access to health services for all citizens, including those with rare diseases. Within the National Health Insurance system (*Jaminan Kesehatan Nasional,* JKN) administered by BPJS Kesehatan, rare diseases are not recognized as a distinct benefits category; under Presidential Regulation No. 59 of 2024, JKN benefits are limited to *Kebutuhan Dasar Kesehatan*, legally defined as "essential needs relating to individual health services for maintaining health, eliminating health disturbances, and saving lives, in accordance with epidemiological patterns and the life cycle," and while the regulation does not explicitly exclude orphan drugs, listings are determined through the National Formulary (Fornas) and health technology assessment by the Indonesian Health Technology Assessment Committee (InaHTAC).[208,209] Furthermore, BPOM Regulation No. 23 of 2025 introduced a definition of Orphan Drug and a 100-day accelerated review for medicines targeting serious and rare conditions, complementing BPOM Regulation No. 24 of 2017; these measures speed authorisation but do not guarantee reimbursement.[210]

Indonesia does not yet operate a publicly documented national rare-disease registry, and available information is dispersed across tertiary hospitals such as Dr Cipto Mangunkusumo Hospital (RSCM), which functions as a de facto referral node for genetic evaluation and research. Evidence from a 2023 peer-reviewed study shows that molecular diagnostics are largely confined to research-based settings because of constrained laboratory capacity, the

208 Ministry of Health Indonesia. (n.d.). *Tentang Fornas*. Kemenkes E-Fornas. Link

209 US Agency for International Development. (2023, November 21). *Strengthening health technology assessment capacity in Indonesia*. The Medicines, Technologies, and Pharmaceutical Services (MTaPs) Programme. Link

210 BPOM. (2025, August 12). *Peraturan Badan Pengawas Obat dan Makanan Nomor 23 Tahun 2025 tentang Perubahan Kelima Atas Peraturan Kepala Badan Pengawas Obat dan Makanan Nomor 24 Tahun 2017 tentang Kriteria dan Tata Laksana Registrasi Obat*. Link

absence of a recognised clinical-genetics service line, and lack of insurance reimbursement for genomic testing; in a rigorously selected cohort of individuals with intellectual disability and multiple congenital anomalies, the study reported a diagnostic yield of 54% using chromosomal microarray and 71% using exome sequencing, demonstrating strong technical potential but also that case-level findings are not yet flowing into a national surveillance or policy data system.[211] The Ministry of Health's Biomedical and Genome Science Initiative (BGSi) is positioned to close these structural gaps by standardizing pipelines for sequencing, biobanking, and clinical data linkage across hospitals, supplying shared data standards, enabling interoperability between screening programmes and tertiary centres, and providing the governance backbone for Indonesia's first national rare-disease registry that can support longitudinal tracking, equitable referral pathways, and evidence-informed benefit decisions under JKN.[212]

Under Presidential Regulation No. 59/2024, JKN benefits are limited to Kebutuhan Dasar Kesehatan and medicine coverage follows Fornas as regulated by Health Minister Decree No. HK.01.07/MENKES/2197/2023 and its amendment No. HK.01.07/MENKES/1818/2024; only medicines listed in Fornas are normally reimbursed, and inclusion depends on InaHTAC evaluations of clinical effectiveness, cost-effectiveness, and budget impact, with a narrow exception allowing referral hospitals to use non-Fornas medicines on a case-by-case basis that are not routinely reimbursed and often place financial responsibility on hospitals. Because JKN uses a tiered referral system with capitation payments at primary care and INA-CBG payments at referral hospitals, patients with rare diseases can generally receive consultations, diagnostics, inpatient care, and supportive services if claims are properly coded and documented, but high-cost orphan drugs are only covered when listed in Fornas, so hospitals rely on non-Fornas approvals, philanthropic donations, research programmes, or manufacturer support, creating unequal access across regions; until InaHTAC evaluations lead to official inclusion in Formularium Nasional, rare disease treatment access remains inconsistent, and Indonesia's rare-disease ecosystem is mainly sustained by family-led and community organisations such as Yayasan MPS dan Penyakit Langka Indonesia which runs fundraising, education, and advocacy activities,[213,214] helped resolve import tax barriers for rare disease medicines and specialised foods in 2016 and continues to mobilise community fundraising, although there is no clear documentation of an institutionalised

211 Sihombing, N. R. B., Winarni, T. I., De Leeuw, N., Van Bon, B., Van Bokhoven, H., & Faradz, S. M. (2023). Genetic diagnostic approach to intellectual disability and multiple congenital anomalies in Indonesia. *Intractable & Rare Diseases Research*, *12*(2), 104–113. Link

212 Puspitasari, W. (2022, August 14). Kemenkes luncurkan BGSi untuk deteksi potensi penyakit di masa depan. *Antara News*. Link

213 Yayasan MPS & Penyakit Langka. (n.d.). *Selamatkan Anak Indonesia dari Penyakit Langka*. Kitabisa.com. Link

214 Juniman, P. T. (2018, March 1). Pemerintah Diminta Masukkan Penyakit Langka dalam BPJS. *CNN Indonesia*. Link

role for patient representatives in agenda setting and policymaking on rare-disease issues.[215]

4.3.2 Malaysia

Malaysia formalised its commitment to rare disease management through the National Policy for Rare Diseases (2025), a major milestone aligning national priorities with World Health Assembly Resolution WHA 78.11 on rare diseases. The policy, endorsed by the Ministry of Health (MOH), outlines nine strategic pillars covering clinical management, orphan medicines, funding and reimbursement, laboratories, registries, advocacy, rehabilitation, and HTA,[216] and envisions an integrated system where PLWRD have equitable access to timely diagnosis, quality treatment, and social support within the broader UHC framework, with the NRDC under MOH providing overall governance and coordination with universities, hospital networks, and patient organisations. Rare disease data collection remains primarily hospital-based, with condition-specific registries managed by tertiary institutions such as Hospital Kuala Lumpur, Universiti Kebangsaan Malaysia Medical Centre (UKMMC), and University of Malaya Medical Centre (UMMC); the Genetic and Metabolic Unit of Hospital Kuala Lumpur maintains records on inborn errors of metabolism and lysosomal storage diseases, and a 2020 study by Shafie et al. identified 1,249 patients across 28 disease categories in public hospitals,[217] prompting the National Policy for Rare Diseases 2025 to outline the establishment of a National Rare Disease Registry coordinated by MOH and the Institute for Medical Research (IMR) to harmonize data standards through ICD-11 and Orphanet coding and integrate genetic and newborn screening information.[218]

Malaysia provides partial public funding for rare-disease care through MOH, mainly via hospital operating budgets and the special medical assistance scheme, with most services centralised in tertiary hospitals such as Hospital Kuala Lumpur, UKMMC, and UMMC that act as referral centres for genetic and metabolic disorders.[219] Only about 60% of patients receive treatment due to high drug costs and limited diagnostic capacity,[220] with enzyme replacement

215 Majelis Permusyawaratan Rakyat. (2023, March 29). *Deteksi Dini Harus Ditingkatkan untuk Pencegahan Penyakit Langka di Tanah Air*. mpr.go.id. Link

216 Ministry of Health Malaysia. (2025). *National Policy for Rare Diseases in Malaysia* (MOH/P/PAK/576.25(GU)-e). Medical Development Division. Link

217 Shafie, A. A., Supian, A., Hassali, M. a. A., Ngu, L., Thong, M., Ayob, H., & Chaiyakunapruk, N. (2020). Rare disease in Malaysia: Challenges and solutions. *PLoS ONE*, *15*(4), e0230850. Link

218 The International Classification of Diseases, 11th Revision (ICD-11) is a tool released by the WHO in 2025 to standardize the language used by health professionals worldwide in diagnosing, reporting, and monitoring diseases, injuries, and causes of death. Orphanet is an international database on rare diseases that provides standardized information for diagnosis, research, and care. It maintains the ORPHAcode system for consistent disease coding and was established in 1997 by the French National Institute for Health and Medical Research (INSERM) with support from the European Commission.

219 Shafie, A. A., Supian, A., Hassali, M. a. A., Ngu, L., Thong, M., Ayob, H., & Chaiyakunapruk, N. (2020). Rare disease in Malaysia: Challenges and solutions. *PLoS ONE*, *15*(4), e0230850. Link

220 Ibid.

therapy (ERT) costing between USD 120,000 (500,000 Malaysian Ringgit) and USD 240,000 (1 million MYR) per patient annually,[221] so access still depends heavily on charitable donations, industry-supported programmes, and out-of-pocket payments; the Rare Disease Trust Fund established in 2022, supported by contributions from individuals, corporations, and civil-society organisations and eligible for income-tax deductions, had raised USD 25,000 (105,000 MYR) by mid-2023 with the Malaysia Lysosomal Diseases Association (MLDA) leading fundraising efforts.[222]

The 2025 national budget allocated an additional USD 5.9 million (25 million MYR) for rare-disease care, primarily to strengthen haemophilia treatment and expand access to cost-effective therapies such as Emicizumab,[223] yet coverage of orphan drugs remains limited, many remain outside the MOH Medicines Formulary and are available only through case-by-case approvals, and insurance coverage is minimal and restricted to a few conditions such as muscular dystrophy and lupus; in this context, advocacy groups including the Malaysian Rare Disorders Society (MRDS), MLDA, and the Malaysia Metabolic Society (MMS), as well as smaller condition-specific groups, have driven awareness campaigns, conferences, and fundraising, with MRDS playing a consultative role in developing the National Policy for Rare Diseases 2025 and MLDA launching the long-term "RM1 for Every Life Counts" campaign in 2016 to sustain ERT and assist patients awaiting treatment.

4.3.3 The Philippines

The Philippines established a legal foundation through the Rare Disease Act of 2016 (Republic Act 10747), the first comprehensive rare-disease law in ASEAN. The Act mandates early diagnosis, access to orphan medicines, and protection of patient rights by classifying PLWRD as persons with disabilities, and directs the Department of Health (DOH) to develop the National Rare Disease Plan and coordinate implementation with the Philippine Health Insurance Corporation (PhilHealth), the Department of Education, and the Department of Social Welfare and Development.[224] The government began strengthening the National Rare Disease Registry in 2017 through a joint effort of the DOH, the University of the Philippines National Institutes of Health (UP-NIH), and the Philippine Society for Orphan Disorders (PSOD), with support from Sanofi Pasteur, aiming to improve identification, diagnosis, and documentation of

221 Rare Disease Malaysia. (n.d.). *About*. rarediseasemalaysia.com. Link
222 Chan, C. S. (2024, January 27). Medical expert says equity in healthcare for rare diseases needed so that no one left behind. *Malay Mail*. Link
223 Malay Mail. (2024, November 5). RM25m boost in rare disease funding may help address haemophilia needs, says Bentong MP Young Syefura. *Malay Mail*. Link
224 Republic of the Philippines. (2016, March 3). *An Act promulgating a comprehensive policy in addressing the needs of persons with rare Disease – RA 10747*. National Council on Disability Affairs. Link

rare diseases;[225] as of January 2017, PSOD had recorded 319 registered patients representing 63 distinct rare disorders.[226]

The law guarantees subsidised care through PhilHealth and DOH hospitals, but actual implementation has been constrained by limited and inconsistent funding. Budget allocation for the Integrated Rare Diseases Management Programme Strategic Plan was first released in 2022 at USD 1.7 million (104.9 million Peso), then drastically reduced to USD 490,000 (28.8 million Peso) in 2023,[227] a level that health policy experts and patient advocates consider insufficient to sustain diagnosis, treatment, and long-term management, with Stratbase President Victor Andres Manhit noting that seven years after the law's passage, patients continue to face disparities in access to medicines and medical specialists.[228] Moreover, established in 2006 with support from UP-NIH and the Institute of Human Genetics, PSOD serves as the cornerstone of patient advocacy and coordination for rare diseases in the country, acting as the national umbrella Organisation for patients, families, and caregivers, leading advocacy, providing patient navigation, and coordinating medical, psychosocial, and financial support services, and playing an instrumental role in drafting and lobbying for the Rare Disease Act of 2016.[229]

4.3.4 Singapore

Singapore advanced its response to rare diseases through the establishment of the Rare Disease Fund (RDF) in 2019, a financing mechanism jointly developed by the Ministry of Health (MOH) and the SingHealth Fund that combines community donations with government matching contributions to support citizens who require high-cost, life-saving medicines that remain unaffordable even after subsidies, insurance, and financial aid such as MediFund. For every USD 0.7 or S$1 donated, the government contributes USD 2.1 (S$3) in matching funds and covers all operational costs so that donations fully benefit patients;[230] managed by an independent committee and guided by clinical experts, the RDF now funds six medicines for four rare diseases, including Gaucher disease, primary bile acid synthesis disorder, Pompe disease, and tetrahydrobiopterin (BH4) deficiency, and with USD 13.8 million (S$18 million) in public donations and USD 53.8 million (S$70 million) in total funds after government matching,

225 Garcia, J. (2017, March 12). *Philippines begins drive to help rare disease sufferers*. Global Genes. Link
226 Philippine Society for Orphan Disorders. (n.d.). *Rare Diseases and the PSOD Rare Disease Registry*. Philippine Society for Orphan Disorders, Inc. Link
227 All currency figures have been converted to USD using the exchange rate published on https://www.xe.com/ as of Sunday, 2 November 2025, when this report was being developed.
228 Melitante, M. (2023, March 30). *Healthcare experts: Rare disease law fund remains 'inadequate.'* inquirer.net. Link
229 Pascual, R. (2016, December 16). *Rare Disease Advocacy takes centre stage at 2016 NBS confab*. Philippine Society for Orphan Disorders, Inc. Link
230 Ministry of Health Singapore. (2019, July 2). *Rare Disease Fund to Provide Financial Support to Singaporeans with Rare Diseases*. Link

it operates as an endowment that uses investment income to sustainably support patients.[231]

While Singapore does not yet have a comprehensive national strategy or legislation for rare diseases, it provides a regulatory framework for access to essential therapies through the Medicines Act (Chapter 176, Section 9) and the Medicines (Orphan Drugs) (Exemption) Order, first issued in 1991 and revised in 2005 and 2016, which allows the import and use of orphan drugs for rare, life-threatening, or severely debilitating conditions affecting fewer than one in 2,000 people; [232] rare disease data are primarily research-driven, with clinical and genetic data maintained at tertiary hospitals such as KKH, National University Hospital (NUH), and Singapore General Hospital (SGH), and supported by the Singapore Rare Disease Models and Mechanisms (RDMM) Network and the Singapore Rare Disease Project (SGRDP) Registry that connect clinicians and scientists working on the same genes and model systems,[233,234,235] laying the foundation for more coordinated data collection and future policy development.[236, 237]

Singapore's funding system for rare diseases is anchored by a mix of public health financing schemes and philanthropic contributions, with patients accessing subsidies through MediShield Life, MediSave, and MediFund, and the RDF serving as an essential safety net for high-cost therapies that exceed these mechanisms.[238] The RDF prioritizes Singapore citizens treated in public healthcare institutions whose treatment costs remain unaffordable even after subsidies, insurance, and financial assistance schemes, operates as an endowment fund using interest income to sustain long-term patient support, and maintains equity and transparency through an independent RDF Committee comprising volunteers, financial experts, and clinicians; Singapore citizens at public healthcare institutions may apply for assistance under the RDF, with clinical and financial eligibility subject to annual review, and as of 22 March 2023 it had supported nine patients, representing about 0.4% of Singaporeans with rare diseases.[239,240] Patient advocacy in Singapore, led by the Rare Disorders

231 Ibid.
232 Attorney-General's Chambers of Singapore. (2005, March 31). *Medicines (Orphan Drugs) (Exemption) Order*. Singapore Statutes Online. Link
233 Ying, L. L. (2024, September 15). Gene detectives: KKH diagnostic programme uncovers rare disorders, helps over 300 families find answers. *KK Women's and Children's Hospital*. Link
234 Singapore General Hospital. (2024, March 22). A One-Stop Genetic Counselling Service for Pregnancy and Pre-conception. *Singapore General Hospital*. Link
235 National University Health System. (2024, April 24). How this paediatrician is helping families tackle rare diseases. *NUHS+*. Link
236 SingHealth. (n.d.). *Singapore Rare Disease Project Registry*. Link
237 SingHealth. (n.d.). *Singapore Rare Disease Models and Mechanisms (RDMM) Network*. Link
238 Ministry of Health Singapore. (2023, March 22). *Singaporeans Diagnosed with Rare Diseases and Coverage by Rare Disease Fund*. Ministry of Health. Link
239 Pearce, F., Lin, L., & Ng, K. (2020). *Funding of treatments for rare diseases in Singapore*. Agency for Care Effectiveness. Link
240 Ministry of Health Singapore. (2023, March 22). *Singaporeans Diagnosed with Rare Diseases and Coverage by Rare Disease Fund*. Ministry of Health. Link

Society Singapore (RDSS) founded in 2011 by parents of affected children,[241] complements this system through financial, emotional, and psychosocial support, including the Medical Intervention Support Scheme (MIS) that offers up to USD 307 (S$400) per beneficiary, the Power for Life (PFL) programme that reimburses up to USD 61.5 (S$80) monthly,[242] a Nutrition Subsidy and a Consumable Support Scheme (CSS), while philanthropic and civil-society partners such as the Tanoto Foundation,[243] Club Rainbow (Singapore), Caring SG, Serving People with Disabilities (SPD), and the Asian Women's Welfare Association (AWWA) contribute funding, counselling, education, rehabilitation, and caregiver support, together creating an integrated support ecosystem for people living with rare diseases.[244]

4.3.5 Thailand

Thailand's approach to rare disease management is anchored in its UHC framework administered through the National Health Security Office (NHSO), which has gradually incorporated rare disease care into national benefit packages and genomic health priorities rather than enacting a single rare disease law. In 2020, the NHSO formally identified 24 rare diseases for inclusion in its benefits scheme and allocated dedicated capitation payments for diagnostic evaluation, confirmatory testing, and lifelong management under specialised centres, marking the first national recognition of rare diseases as a public health priority; [245] between 2021 and 2022, the NHSO expanded the NLEM under category E2, which covers high-cost medicines requiring specialist supervision, to include 14 orphan drugs,[246,247] enabling centralized procurement and equitable distribution through designated referral hospitals,[248] supported by economic evaluations from the HITAP that recommended newborn screening and early treatment for selected inborn errors of metabolism (IEMs) as cost-effective.[249] In parallel, the country launched Genomics Thailand, a national precision medicine initiative under the Ministry of Public Health, and established the Thailand Rare and Undiagnosed Disease Network (T-RUN), composed of multiple medical centres to provide a collaborative platform for genomic sequencing, patient referral, and policy coordination, positioning

241 SG Enable. (n.d.). *Rare Disorders Society (Singapore)*. Enabling Guide. Link
242 Rare Disorders Society Singapore. (n.d.). *Programmes & Services*. RDSS. Link
243 Tanoto Foundation. (2022, June 28). *Tanoto Foundation donates to Rare Disease Fund in Singapore*. Link
244 Rare Disorders Society Singapore. (n.d.). *Seek Help*. RDSS. Link
245 National Health Security Office. (2021b, March 2). *24 rare diseases added to UCS But challenges remain*. NHSO. Link
246 National Health Security Office. (2021, August 25). *Four orphan drugs added to the national drug list*. NHSO. Link
247 National Health Security Office. (2022, July 28). *10 orphan drugs added to the national drug list*. NHSO. Link
248 The designated hospitals, mostly located in Bangkok, include Chulalongkorn Hospital, Thammasat Hospital, Siriraj Hospital, Ramathibodi Hospital, Phramongkutklao Hospital, Queen Sirikit National Institute of Child Health, and Srinagarind Hospital in Khon Kaen.
249 Wichajarn, K., Sawatjui, N., Prasongdee, P., Panklin, A., Sornkayasit, K., Chungkanchana, N., Tessiri, S., Wintachai, P., Dechyotin, S., Pasomboon, C., Ratanapontee, J., Thanakitsuwan, S., & Rattanathongkom, A. (2025). The Establishment of Expanded Newborn Screening in Rural Areas of a Developing Country: A Model from Health Regions 7 and 8 in Thailand. *International Journal of Neonatal Screening*, *11*(2), 26. Link

Thailand as one of the few middle-income countries in the region integrating genomics and rare disease care within the UHC system.[250]

Thailand has progressively developed data systems to support rare disease policy and surveillance, including the Thailand Birth Defects Registry established in 2012 by the Queen Sirikit National Institute of Child Health (QSNICH) in collaboration with the NHSO and the Ministry of Interior,[251] which integrates four main data sources (birth registration, NHSO reimbursement data, hospital reports, and an online reporting platform) linked by the 13-digit national identification number and ICD-10 codes and which recorded 67,813 live births and 3,696 congenital anomalies in its first year, a prevalence of 26.12 per 1,000 live births.[252] A major advancement occurred in 2022 when the Expanded Newborn Screening (NBS) Programme was scaled up nationwide to cover all 13 health regions, using tandem mass spectrometry to screen for congenital hypothyroidism and 40 inborn errors of metabolism; the pilot in Health Regions 7 and 8 achieved 98.6% coverage of 123,692 live births, identified 101 congenital hypothyroidism and 20 IEM cases, and reduced reporting time from 9.1 to 8.4 days through digital workflow integration,[253] using a real-time data platform (KKU-IEM) for sample tracking and electronic reporting.

Genomics Thailand has further expanded diagnostic capacity by introducing exome sequencing as a first-tier test for undiagnosed or rare conditions through the T-RUN network of tertiary hospitals and provides the groundwork for a future National Rare Disease Registry linking genetic, clinical, and newborn screening data; rare disease services are primarily funded through the UCS, NLEM-listed treatments are fully reimbursed by the NHSO, and delivery is organised through seven regional rare disease centres, six in Bangkok and one at Srinagarind Hospital, Khon Kaen University, with annual funding per patient ranging from approximately USD 1,515 to 9,091.[254] However, only 46.8% of medicines recommended by the International Rare Diseases Research Consortium (IRDiRC) are registered in Thailand, just 22.93% are listed in the NLEM, and only 31.7% were procured for hospital use over the past five years,[255] revealing persistent access barriers, while patient advocacy led by the Thai Rare

250 Shotelersuk, Vorasuk. (n.d.). *Genomics Thailand: Rare Diseases* [Slide show]. Health Systems Research Institute (HSRI). Link

251 Pangkanon, S., Sawasdivorn, S., Kuptanon, C., Chotigeat, U., & Vandepitte, W. (2014). Establishing of National Birth Defects Registry in Thailand. *Journal of the Medical Association of Thailand = Chotmaihet thangphaet*, *97 Suppl 6*, S182–S188.

252 Ibid.

253 Wichajarn, K., Sawatjui, N., Prasongdee, P., Panklin, A., Sornkayasit, K., Chungkanchana, N., Tessiri, S., Wintachai, P., Dechyotin, S., Pasomboon, C., Ratanapontee, J., Thanakitsuwan, S., & Rattanathongkom, A. (2025b). The Establishment of Expanded Newborn Screening in Rural Areas of a Developing Country: A Model from Health Regions 7 and 8 in Thailand. *International Journal of Neonatal Screening*, *11*(2), 26. Link

254 Ibid.

255 Suwattanapreeda, S., Hirunrassamee, S., Sooksriwong, C., Maluangnon, K., Chuachantra, T., Kuchaisit, K., & Osirisakul, N. (2025). Unlocking access: a comprehensive analysis of medicines accessibility for rare diseases in Thailand. *Orphanet Journal of Rare Diseases*, *20*(1). Link

Disease Foundation (TRDF) and groups such as NF Club Thailand has become essential for awareness, family support, and collaboration with the NHSO to expand treatment coverage, strengthen referral networks, and improve data systems.[256,257]

4.3.6 Viet Nam

Viet Nam has not enacted a dedicated rare disease law, but in recent years the Ministry of Health (MoH) has elevated rare disease as a policy priority through institutional mechanisms, including the establishment of a Steering Committee for Strengthening Rare Disease Management led by the Deputy Minister and engaging agencies, academic societies, and patient stakeholders.[258] In 2025, revisions to the Health Insurance Law (Law No. 51/2024/QH15) were passed to allow patients with 62 designated rare or severe diseases to access 100% insurance coverage even when seeking care in higher-level hospitals without a referral document,[259,260] a measure intended to remove administrative barriers and accelerate access, although coverage remains incomplete because certain rare conditions and orphan drugs are excluded from standard benefits and some patient groups such as older patients with late-diagnosed conditions still bear high out-of-pocket costs;[261] the government is also exploring regulatory flexibility to encourage orphan drug availability, and under Circular 26/2019/TT-BYT has introduced regulatory incentives including fast-track approval, simplified import procedures, and fee exemptions for orphan drugs for rare disease patients.

Viet Nam currently does not maintain a unified nationwide rare disease registry or integrated surveillance system; instead, clinical centres and specialist hospitals maintain isolated case registers that are seldom harmonised or shared across institutions, and national estimates suggest about 6 million people in the country live with rare diseases, with 58% being children, while many cases remain undiagnosed or misdiagnosed and the true burden is uncertain.262 Some regulatory tools support data collection, since Circular 26/2019/TT-BYT defines a listing process for orphan drugs that requires evaluating epidemiological evidence and disease prevalence, and genomic

256 The Nation. (2021, March 12). 24 rare diseases included in universal coverage, but challenges remain. *Thenation*. Link
257 AztraZeneca. (2024, June 14). *Empowering Survivors: The "BORN a FIGHTER" campaign supports and educates neurofibromatosis (NF) warriors, celebrating the 7th event on NF1 Awareness Day in Thailand*. Link
258 *Việt Nam faces limited access to treatment for rare diseases*. (2025, August 20). Viet Namnews.vn. Link
259 Viet Nam Social Security. (2025, March 14). *Universal Health Insurance – A Chance for Equal Access to Healthcare Services*. The Ministry of Finance. Link
260 Thị, B. K. T. V. Đ. (2025, January 2). 62 rare and dangerous diseases do not require referral papers and are still entitled to 100% health insurance. *Viet Nam.vn*. Link
261 Son, T. (2024, March 5). Supportive policies needed to give rare diseases people more opportunities. *SGGP English Edition*. Link
262 Nguyen, K. (2024, March 1). Six million Vietnamese people with rare diseases difficult to access drugs. *SGGP English Edition*. Link

testing capacity is growing in tertiary hospitals through research collaborations and pilot projects, although there is still no national database linking genomic data to disease surveillance and policy decisions.[263]

Funding for rare disease care is mediated primarily through the national health insurance system (Viet Nam Social Security or VSS) plus hospital budgets, manufacturer assistance, philanthropy, and pilot programmes; treatment for the 62 designated diseases is now eligible for 100% insurance reimbursement and the 2025 reform removes the requirement for referral letters for rare, severe, or high-technology conditions, yet not all orphan drugs or advanced therapies will be covered fully, and service delivery remains concentrated in major provincial and central referral hospitals where limited specialist capacity and geographic barriers mean many patients face long travel, delays, fragmented care, and out-of-pocket payments. Patient advocacy has strengthened in recent years through the Vietnamese Organisation for Rare Diseases (VORD), a non-profit social enterprise established in 2021 that focuses on spinal muscular atrophy, Duchenne muscular dystrophy, brittle bone disease, and other rare conditions, with a mission to raise awareness, improve diagnosis and treatment, build patient communities, connect local groups with global networks, and provide information and training through VORD Academy to help patients access medical equipment, develop new skills, find employment, and in some cases access treatment abroad.[264]

263 Ministry of Health Viet Nam. (2019). *Circular No. 26/2019/TT-BYT on List of Orphan Drugs*. Socialist Republic of Viet Nam. Link

264 Do, H. P. (n.d.). *Vietnamese Organisation for Rare Diseases*. SMA Europe. Link

ANNEXURE 4.

Summary of Breakout Session

The breakout discussions across the four groups revealed common priorities for ASEAN, including strengthening policy frameworks, diagnostic and genomic capacity, referral pathways, and treatment access through improved governance, cross-country collaboration, and workforce development. Participants highlighted persistent delays in diagnosis, uneven access to treatment, fragmented regulatory and financing mechanisms, and heavy reliance on charity or ad hoc support. They called for stronger registries, resource-sharing, more flexible regulatory pathways, expanded insurance coverage, innovative financing models, and deeper engagement with patient organisations and industry. Overall, the sessions underscored a shared regional commitment to building more coordinated, sustainable, and equitable systems for rare-disease diagnosis, care, and treatment

The breakout session on *Translating the WHA Resolution into ASEAN Realities* highlighted shared priorities across Member States, including the need for stronger policy frameworks, improved regional and national collaboration mechanisms, and enhanced diagnostic and genomic capacity. Participants underscored the importance of developing national and regional registries, sharing resources for testing facilities, and integrating rare diseases into primary care and public-health programmes. Workforce gaps across clinical, laboratory, and allied health cadres prompted calls for expanded training, specialist development, and cross-country exchanges. Financing challenges were also evident, with countries seeking sustainable funding models, insurance inclusion, and mechanisms such as public–private partnerships and pooled procurement. Collectively, the discussion reflected a clear regional mandate to strengthen governance, workforce capability, and financing arrangements to translate global commitments into practical national action.

Table 10. Summary of Breakout Session about Translating the WHA Resolution into ASEAN Realities

Topics	Domain	Country	Key Points
Infrastructure	Government Structures	Malaysia	Proposal to develop an ASEAN regional framework for rare disease control.
		Viet Nam	National plan supported by a network of experts, patients, and families.
	Policy Frameworks	Malaysia	Suggested advancing an ASEAN framework for rare disease governance.
	Collaboration Platforms	Indonesia	Encouraged regional research collaboration on rare diseases.
		Malaysia	Noted limited collaboration with the private sector.
		Singapore	Proposed strengthening ASEAN collaboration on screening, confirmation, management, and treatment.
	Digital Platforms (registries, data sharing)	Indonesia	Proposed standardising a rare disease registry at regional level.
		Malaysia	Emphasised development of national registries.
		Singapore	Suggested establishing a national rare disease registry.
	Testing Facilities	Malaysia	Proposed sharing regional resources for molecular and genomic testing.
		Singapore	Highlighted need to incorporate screening for rare diseases.
		Thailand	Raised long timelines and capacity needs for diagnostics.
	Treatment Delivery Systems	Indonesia	Proposed strengthening regional access to food and medicines for rare diseases.
		Malaysia	Suggested integrating rare diseases into primary care and broader clinical services.
		Thailand	Prioritised equitable access to essential diagnostics through the National Essential Drug List.
	Legislation	Malaysia	Noted absence of dedicated rare disease legislation.
	Other Issues	Malaysia	Limited support for families, including financial, rehabilitation, and community care.
		Viet Nam	Highlighted the role of media in raising awareness among policymakers.

Topics	Domain	Country	Key Points
Manpower	Clinicians	Indonesia	Proposed expanding training for health workers managing rare diseases.
		Malaysia	Called for structured training for geneticists, scientists, and clinicians.
		Singapore	Identified need for experts and centres of excellence.
		Viet Nam	Referred to national guidelines for rare disease care.
	Allied Health Professionals	Indonesia	Encouraged shared training and capacity building.
		Malaysia	Identified shortages in laboratory personnel, genomic officers, and counsellors.
		Philippines	Suggested developing pre-service curricula for allied health workers.
		Singapore	Proposed exchanges of experts and technicians.
		Viet Nam	Highlighted potential for national and ASEAN collaboration.
	Integration into UHC	Indonesia	Noted that heavy subsidies for rare disease medicines are not sustainable.
		Philippines	Need for financial mechanisms, including insurance.
		Thailand	Proposed economic studies to support UHC expansion for rare diseases.
		Viet Nam	Government coverage is partial; external support needed.
	Public–Private Partnerships	Indonesia	Raised pooled procurement due to high treatment costs.
	National Insurance Schemes	Indonesia	Rare diseases are not yet covered.
		Malaysia	Proposed establishing an ASEAN trust fund for regional support.
		Philippines	Need for creative financing mechanisms.
		Thailand	Suggested that countries establish funds to support high-cost diagnostics and treatment.
		Viet Nam	Highlighted increasing healthcare costs due to prolonged cases.
	Other Issues	All Countries	Recognised the need for sustainable and innovative financing models.

The discussion on *Patient Journey from Diagnosis to Care* revealed persistent delays in early diagnosis across ASEAN, driven by limited provider awareness, inconsistent definitions of rare diseases, and late identification of conditions for which treatment exists. Countries emphasised the need for capacity building, advanced diagnostic tools, and partnerships with industry and international institutions. Strengthening referral pathways emerged as a critical priority, including the use of precision-medicine tools, wider patient-organisation networks, digital health solutions, and cross-border collaboration to reduce delays in reaching specialist care. Continuity of care remains uneven, with participants highlighting the roles of patient organisations, free sequencing support, multi-stakeholder collaboration, and structured private-sector engagement. Overall, countries aligned on the need for stronger networks, enhanced capacity, and integrated pathways that link early diagnosis to long-term care.

Table 11. Summary of Breakout Session about Patient Journey from Diagnosis to Care

Domain	Country	Key Points
Early Diagnosis	Indonesia	1. Major gap in health care providers' ability to identify and diagnose rare diseases. Need to prioritise treatable conditions in policy. 2. Need support for capacity building, analysis, and regional cooperation; tech transfer and overseas training recommended.
	Malaysia	Partnership with industry is a good option.
	Philippines	Late diagnosis of NF1, often at an advanced age.
	Viet Nam	Differences in the definition of rare diseases.
Referral Pathways	Singapore	Precision-medicine tools can support early detection (e.g. facial-recognition tools). Opportunities for capacity-building and industry partnership in training and tool development.
	Thailand	Need to build more extensive networks of patient-organization leaders across the region.
	Viet Nam	Outreach and capacity building for early diagnosis; use of digital health and teleconsultation to shorten pathways, especially for patients living far from centres of excellence.
Continuity of Care	Indonesia	Engagement with CDLS organisation; free sequencing support for patients and families to overcome cost barriers.
	Philippines	Need collaboration among stakeholders to strengthen referral pathways, diagnosis, and continuity of care.
	Singapore	Private sector is an important partner; need to align opportunities and patient readiness. Singapore has guidelines for collaboration with industry.

The breakout session on *Equitable Pathways to Treatment* underscored significant gaps in regulatory processes, financing, regional collaboration, data systems, patient engagement, and capacity building. Countries reported slow or fragmented regulatory pathways, limited or unsustainable financing mechanisms, and continued reliance on international donations. Participants emphasised the need for more flexible regulatory approaches, dedicated funding mechanisms, trust funds, and structured public–private partnerships to secure sustainable access to treatment. Strengthened regional collaboration, improved data systems to support HTA and policy development, and deeper engagement with patient organisations were also identified as essential. Differences across Member States highlighted the need for practical capacity-building programmes for clinicians and institutions to support earlier diagnosis, efficient referral, and consistent long-term management. Ensuring equitable access will require a coordinated approach combining regulatory reform, innovative financing, stronger data infrastructure, and cohesive multi-stakeholder action.

Table 12. Summary of Breakout Session about Equitable Pathways to Treatment

Domain	Country	Key Points
Regulations	Indonesia	1. HTA Committee exists but is reviewing other health topics beyond rare diseases. 2. A new submission scheme is being developed so researchers and academia can propose priority programmes for HTA review. 3. Proposals with supporting data can be analysed for potential inclusion in national priorities.
	Malaysia	1. Management guideline simplifies approval for orphan drugs, but the approval process is slow. 2. Malaysia has approved more than 20 rare-disease products. 3. Lack of financial mechanisms means approved drugs often cannot be procured.
	Philippines	1. Philippines has a Rare Disease Act, but it is not fully funded. 2. Definition of rare disease still evolving. 3. Advocacy ongoing to expand government funding and integrate rare diseases into national health priorities.
	Singapore	1. Highlighted importance of a collaborative framework across partners to strengthen rare-disease policies. 2. Emphasised that priorities must reflect different country needs and capacities. 3. Tailored approaches are needed to achieve equitable progress.
	Thailand	1. Treatment access remains limited due to insufficient financing. 2. Most therapies are not reimbursed under national insurance, forcing reliance on international donations. 3. Only about 30% of patients over six years old are covered under existing schemes.

Domain	Country	Key Points
Financing	Malaysia	1. Dedicated annual allocation exists, but funding is insufficient. 2. Rare diseases require lifelong care, creating ongoing gaps. 3. Malaysia is exploring trust funds and partnerships to ensure sustainable access.
	Philippines	1. Very limited financing; only few treatments are included in government programmes. 2. High-cost treatments are not covered, creating access gaps. 3. Advocacy continues to increase budget allocations.
	Thailand	1. Rare diseases receive limited funding. 2. Advocacy is important to raise awareness and secure budgets. 3. Even when medicines are available, financial constraints hinder purchase.
Regional Collaboration	Malaysia	Collaboration is easier when involving academia.
Data Infrastructures	Philippines	1. Data on patient journeys is limited. 2. Government and partners are working to build systems and evidence collection. 3. Will support HTA processes and evidence-based policy.
	Thailand	1. NGS diagnostics remain costly. Efforts ongoing to find more affordable alternatives. 2. Exploring partnerships for specific rare-disease groups. 3. Many rare-disease cases require multiple therapies. 3. Pharmaceutical companies find orphan-drug production costly due to low demand. 4. National health benefit coverage limited to special cases.
Patient and Industry Engagement	Malaysia	Patients can access treatment even when drugs are not registered; drug registration is not a major barrier.
	Thailand	Registry interventions can use regional pathways; early industry engagement helps accelerate treatment access.
Capacity Building	Malaysia	1. Need to strengthen physician capacity for diagnosing and managing rare diseases. 2. Both short-term and long-term training needed. 3. Stronger expertise will improve early detection, accurate diagnosis, and patient care.

The discussion on *Sustainable and Innovative Financing* showed that most ASEAN countries currently rely on fragmented or ad hoc approaches, including charity partnerships, limited insurance coverage, crowdfunding, and small government funds. Fiscal contributions and levies remain under-utilised, while social health insurance benefits are uneven and often insufficient. Participants proposed a range of potential financing mechanisms, including earmarked taxes, expanded insurance coverage, industry-based levies, genetic non-discrimination frameworks, and multiparty contribution models such as Singapore's 3-to-1 fund. Stronger partnerships with industry were also identified

as an important enabler. Across the region, there was clear recognition of the need for predictable, pooled, and sustainable financing models that reduce reliance on external donations and ensure equitable access to diagnostics and treatment.

Table 13. Summary of Breakout Session about Sustainable and Innovative Financing

Domain	Current Funding Sources (Countries & Inputs)	Potential Financing Models to Expand Access (Countries & Inputs)
Levies	**Malaysia:** 25 million	**Indonesia:** orphan drugs
Social health insurance schemes	· **Malaysia:** RM8,000 one-off treatment (MySalam) · **Philippines:** treatment for IEM · **Singapore:** coverage available but likely insufficient · **Thailand:** screening, testing, and treatment for IEM · **Viet Nam:** IEM	· **Malaysia:** Genetic Information Non-Discrimination Act, PERKESO · **Philippines:** expand beyond newborn screening · **Thailand:** feasible for RD · **Viet Nam:** screening, diagnosis, treatment, and prevention
Health impact funds	–	**Malaysia:** Social Exchange Impact Fund
Charity partnerships	· **Indonesia:** pharma (compassionate access) · **Malaysia:** pharma + NGOs + teaching hospitals · **Philippines:** pharma + GLCs + NGOs · **Thailand:** pharma + patient fund-raising · **Viet Nam:** pharma + patient fund-raising	–
Multiparty contribution mechanisms	· **Indonesia:** crowdfunding for diagnosis and testing · **Malaysia:** government RD fund + crowdfunding · **Singapore:** 3-to-1 RD fund (9 conditions, 13 treatments) · **Viet Nam:** industry sponsor + patient payment	**Malaysia:** similar to Singapore 3-to-1 model **Philippines:** similar to Singapore 3-to-1 model **Viet Nam:** co-payment
Public–private partnerships	**Philippines:** projects led by patient groups	**Malaysia:** with pharma
Fiscal contributions (tax-based)	–	**Indonesia:** tobacco **Malaysia:** tobacco, alcohol, sugar, gaming **Philippines:** tobacco, alcohol, sugar

ASEAN countries share common gaps in diagnosis, access to treatment, financing, and governance, underscoring the need for stronger collaboration, clearer policies, and more sustainable systems for rare-disease management. Across all groups, participants emphasised improving diagnostic capacity, expanding financing mechanisms, and engaging patients, industry, and regional partners to achieve more equitable and coordinated care.

ANNEXURE 5.

National Priorities and Timelines for Rare Disease Management

Table 14. National Priorities and Timelines for Rare Disease Management

Country	Short Term	Medium Term	Long Term
Indonesia	1. Pilot expanded newborn screening in Jakarta and other high-prevalence areas. 2. Build capacity among healthcare professionals, including clinical geneticists and bioinformaticians, to support the identification and management of rare diseases. 3. Include patient perspectives in developing the national list of rare diseases. 4. Conduct multi-stakeholder consultations to build alignment and establish a national Rare Disease committee under the Ministry of Health.	1. Strengthen funding and outsourcing mechanisms to expand access to diagnostic testing. 2. Establish specialised hubs or centres of excellence within the ASEAN region. 3. Explore Thailand's managed entry agreement system as a potential model for access to rare disease treatments. 4. Promote public co-funding mechanisms to support rare disease diagnosis and treatment. 5. Leverage corporate social responsibility funding, drawing on approaches used in stunting and other public health initiatives. 6. Explore faith-based funding mechanisms as complementary sources of support. 7. Develop a national rare disease registry.	1. Include genomic diagnostics, including next-generation sequencing, and selected rare disease treatments within the BPJS scheme, supported by the development of standardised clinical guidelines.
Malaysia	1. Capacity building 2. Clinical Advisory Group to establish transparent review mechanisms for disease inclusion and resource prioritisation (testing and treatment). 3. National registry 4. Targeted newborn screening.	1. Multi-party Advisory Group, including patient advocacy groups. 2. Capacity building. 3. Expanded newborn screening 4. Centres of Excellence	1. Legal protection against genetic discrimination. 2. Capacity building.

The Philippines	1. Place patients and families at the centre of all efforts, from planning through implementation and evaluation. 2. Increase awareness and empower communities to serve as strong voices for people living with rare diseases. 3. Strengthen psychosocial support for patients and families, particularly those caring for children with rare diseases. 4. Establish a national rare disease registry. 5. Develop clinical practice guidelines to define standards of care for rare diseases. 6. Strengthen HTA capacity to better understand rare disease treatment pathways. 7. Increase awareness among all stakeholders. 8. Encourage clinical trials to support research and innovation in rare diseases. 9. Establish a dedicated rare disease fund, through public, private, or blended financing mechanisms. 10. Explore faith-based funding mechanisms to support rare disease initiatives.	1. Establish centres of excellence for rare diseases. 2. Establish a national rare disease registry. 3. Develop clinical practice guidelines to define standards of care for rare diseases. 4. Increase awareness among all stakeholders. 5. Encourage clinical trials to support rare disease research and innovation. 6. Establish a dedicated rare disease fund, either public, private, or blended. 7. Explore faith-based funding mechanisms to support rare disease initiatives.	1. Encourage clinical trials to support rare disease. 2. Establish a Dedicated Rare disease fund (either public or private).
Singapore	1. Establish a national rare disease registry. 2. Conduct quality of life (QoL) surveys 3. Strengthen regional coordination on drug registration.	1. Develop sustainable financing mechanisms, including blended funding models involving philanthropy and faith-based sources.	1. Increase awareness and education on rare diseases, including carrier screening. 2. Engage with insurance providers to prevent discrimination against individuals diagnosed pre-symptomatically.

Thailand	1. Develop a budget allocation plan. 2. Develop a national roadmap with clear objectives and achievable milestones. 3. Collaborate with industry on co-payment models for orphan drugs. 4. Initiate the development of a unified rare disease registry. 5. Conduct research on the quality of life of patients and their families. 6. Develop genomic data protection policies and regulations.	1. Update and expand the budget allocation plan. 2. Scale up the unified rare disease registry. 3. Improve access to genomic testing, making services more affordable and available nationwide. 4. Strengthen collaboration among ASEAN Member States on genetic testing facilities, HTA, and bioinformatics. 5. Build regional capacity by training genetic counsellors, clinical geneticists, and related professionals. 6. Integrate rare disease data into a national electronic health record system. 7. Develop co-funding models for rare disease diagnosis and treatment.	1. Sustain and institutionalise budget allocation mechanisms. 2. Establish and enforce non-discriminatory act.
Viet Nam	1. Establish national rare disease registry. 2. Strengthen referral and teleconsultation systems. 3. Ensure equitable capacity building among medical workers.	1. Strengthen stakeholder collaboration across sectors. 2. Integrate genetic counselling in care pathways. 3. Use national data to advocate for sustained funding. 4. Include orphan drugs in national priority lists. 5. Assess long-term effectiveness of therapies through data and research.	1. Develop sustainable financing and reimbursement policy mechanisms. 2. Facilitate clinical trials and research collaborations. 3. Reform regulations to improve medicine access.

ANNEXURE 6.

List of the Participants of the Southeast Asia Rare Disease Policy Forum: Advancing Regional Collaboration to Improve the Lives of the Peoplewith Rare Conditions in ASEAN Region

No	Name	Designation	Institution
MALAYSIA			
1	Datuk Seri Dr. Dzulkefly bin Ahmad	Minister of Health	Ministry of Health Malaysia
2	Datuk Dr. Mahathar bin Abd Wahab	Director General of Health	Ministry of Health Malaysia
3	Datuk Dr. Nor Fariza binti Ngah	Deputy Director General of Health (Research & Technical Support)	Ministry of Health Malaysia
4	Dato' Indera Dr. Nor Azimi binti Yunus	Deputy Director General of Health (Medical)	Ministry of Health Malaysia
5	Dr. Azuana binti Ramli	Deputy Director General of Health (Pharmaceutical Services)	Ministry of Health Malaysia
6	Dato' Dr. Mohd Azman bin Yacob	Director	Medical Development Division Ministry of Health Malaysia
7	Dr. Adiratna binti Mat Ripen	Head of Cancer Research Centre	Institute for Medical Research, National Institutes of Health Ministry of Health Malaysia
8	Dr. Julaina binti Abdul Jalil	Head of IEM & Genetics Unit	Institute for Medical Research, National Institutes of Health Ministry of Health Malaysia
9	Dr. Ngu Lock Hock	Head of the Department of Genetics	Hospital Kuala Lumpur (HKL) Ministry of Health Malaysia
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11	Dr. Sangeeta Subramaniam	Senior Principal Assistant Director	Medical Development Division Ministry of Health Malaysia
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17	Dr. Ainul Nadziha binti Mohd Hanafiah	Deputy Divisional Secretary	Division of Policy and International Relations Ministry of Health Malaysia
18	Dr. Tee Khim Boon	Senior Assistant Director	National Pharmaceutical Regulatory Agency (NPRA) Ministry of Health Malaysia
19	Dr. Ami Fazlin binti Syed Mohamed	Director	Institute for Medical Research, National Institutes of Health Ministry of Health Malaysia
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21	Dr. Tan Lu Ping	Head of Molecular Pathology Unit	Institute for Medical Research, National Institutes of Health Ministry of Health Malaysia

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26	Prof. Dr. Thong Meow Keong	Clinical Geneticist	Universiti Malaya Medical Centre
27	Prof. Dr. Lim Soo Kun	Professor and Consultant Nephrologist	Universiti Malaya
28	Prof. Datin Dr. Norlinah binti Mohamed Ibrahim	Professor and Consultant Neurologist	Deputy Dean, Faculty of Medicine Universiti KebangsaanMalaysia
29	Bruce Lim	Interim member & President	Rare Disease Coalition Malaysia (RDCM) & Malaysian Patients Organisation for Primary Immunodeficiency (MyPOPI)
30	Nadiah Hanim binti Abdul Latif	President	Malaysia Rare Disease Society Malaysia (MRDS)
31	Shakira binti Jamil binti Fisal	President	Spinal Muscular Atrophy Malaysia (SMAM)
32	Saida binti Abu Bakar	Interim member & President	Rare Disease Coalition Malaysia (RDCM) & Dystrophic Epidermolysis Bullosa Research Association (DEBRA) Malaysia

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38	Dr. Isra Sabrina	Official	National Biomedical & Health Genomics Center, MoH
39	Dr. Chairunnisa	Official	National Biomedical & Health Genomics Center, MoH
40	Dr. Klara Yuliarti	Pediatrician	Cipto MangunkusumoHospital / Human Genetic Research Centre IMERI
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44	Dr. Chin Hui-Lin	Senior Consultant	National University Hospital
45	Gregory Vijayendran	Chairman	Rare Disease Fund
46	Phua Wee Seng	Executive Director and Chief Rare Advocate	Rare Diseases Society Singapore

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50	Prof. Duangrurde Wattanasirichaigoon	Professor, Clinical Geneticist	Faculty of Medicine Ramathibodi Hospital, Mahidol University
51	Dr. Wuttichart Kamolvisit, MD, MSc (Dist), ECMGG	Pediatrician and Clinical geneticist	King Chulalongkorn Memorial Hospital
52	Dr. Chulaluck Kuptanon	Clinical Geneticist	Department of Medical Service, Ministry of Public Health, Thailand
THE PHILIPPINES			
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55	Ms. Araceli Lanorio	Lead Advocate	Neurofibromatosis Friends (NF Friends)
56	Dr. Loudella V. Calotes-Castillo	Clinical Associate Professor	Rare Disease Special Interest Group, Philippine Neurological Association
57	Dr. Carmencita David Padilla, MD, MAHPS	Professor of College of Medicine	University of the Philippines Manila
VIET NAM			
58	Dr. Nguyen Ngoc Khanh	Deputy Director and Clinician	Center for Endocrinology - Metabolism - Genetics - Molecular Therapy, National Children's Hospital
59	Huy Phuoc Do, MD, MSc.	CEO & Co-Founder	Vietnamese Organization for Rare Diseases (VORD)
60	Dr. Nguyen Thi Thanh Huong	Deputy Director	Ho Chi Minh City PediatricHospital 1

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