

## Malaysia's Evolving Rare Disease Ecosystem: Challenges, Interventions & System Imperatives

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### Introduction

Rare diseases (RDs) collectively constitute a significant yet often overlooked component of national public health systems. The World Health Organization (WHO) defines a rare disease (RD) as any disease which affects a small percentage of the general population.<sup>1</sup> EURORDIS (European Alliance of Rare Disease Patient Organizations) defines RDs as conditions affecting 5 in 10,000 of the general population<sup>2</sup> while in Malaysia, it was first recognised as a disease entity with the opening of the first genetic service in Universiti Malaya Medical Centre (UMMC) in 1995. A modification made was to amend the definition of RD as a condition with a prevalence of 1 in 4,000 of the general population (<http://www.mrds.org.my>) which was subsequently adopted by Ministry of Health (MOH) Malaysia in 2018/2019.<sup>3</sup> As of March 2023, the MOH has identified nearly 500 such RD conditions nationwide.<sup>4</sup> Although each RD is individually rare, collectively they number over 6,000 types and the RD community makes up 3.5 – 5.9% of world population. RDs may happen to anyone regardless of race, gender, age, or socio-economic background. Over seventy percent of RD are genetic in origin; and 30% of the patients passed away by the age of five years, unfortunately.<sup>5</sup> The survivors with RDs often need long term medical care. RDs are non-communicable diseases (NCDs) but due to the lack of registry, there is little data on their epidemiology in Malaysia. The United Nations General Assembly in 2021 approved a resolution on addressing the

challenges of persons living with a RD and their families.<sup>6</sup> Yet, patients with RDs face stigmatisation and arduous “diagnostic odysseys” – delayed treatment due to late diagnosis, low awareness by members of the public and healthcare professionals, limited access to genetic services and reduced access to life-saving medicines.

For families with children or adults living with RDs, the healthcare system must provide lifelong and comprehensive support. This includes early detection, continuous clinical management, access to specialised medicines and therapeutic interventions, and psychosocial assistance for patients and caregivers. A holistic and life-course approach is essential to ensure equitable quality of life and to prevent undue emotional, financial, and logistical burdens on families. The ecosystem surrounding RD care comprising geneticists, genetic counsellors, diagnostic laboratories, rehabilitation services, non-governmental organisations (NGOs), and patient support groups, is inherently interconnected. Each component plays a vital role in facilitating early diagnosis, treatment continuity, emotional well-being, and social inclusion.

A central driver enabling this ecosystem to function effectively is sustainable and continuous financing. Without predictable funding for diagnostics, medicines, rehabilitative services, and community-based support systems, RD patients risk falling through systemic gaps. Strengthened awareness

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across the general public, health professionals, and policymakers at national, regional, and global levels, is also indispensable. Crucially, societal perceptions of the RD community must evolve. RD patients should be regarded not as passive recipients of welfare but as citizens entitled to equitable access to healthcare, education, employment, and dignified living conditions. With increased visibility and resources, Malaysia implemented key transformative strategies, such as developing a national RD registry, enhancing inter-ministerial coordination, and empowering patient advocacy networks to support individuals from birth to end-of-life culminating in a landmark National Policy on Rare Diseases launch in August 2025.<sup>7</sup>

### **Key Challenges Faced by Rare Disease Patients and Families**

Although Orphanet's 2020 estimates suggest that 3.5 – 5.9% of the global population may be affected by an RD,<sup>5</sup> equivalent to at least one million Malaysians affected, public perception continues to frame RD as a marginal issue. This contrasts sharply with the attention given to more common NCDs, such as cardiovascular diseases, diabetes mellitus, and cancer. Given their high population burden and chronic, debilitating nature, RDs should be conceptualised as a major NCD subgroup within national health planning.

The longstanding neglect of RD in public health policy stems partly from insufficient knowledge and awareness among healthcare professionals. Malaysia faces a significant shortage of geneticists, with only

15 to 18 specialists serving the entire country, and genetic counselling remains unrecognised in the public service scheme. This limits the availability of early diagnostic services and contributes to delayed or inappropriate clinical management. Moreover, genetic clinics are not present in every state, creating geographical inequities in access.

Financial constraints further exacerbate these challenges. National RD funding remains inadequate and is often restricted to known patients, leaving limited provisions for new diagnoses or family support. Expanded newborn screening is still not implemented nationwide in Malaysia. While conditions such as inborn errors of metabolism, primary immunodeficiencies, endocrine disorders, and lysosomal storage diseases can often be treated effectively if detected early, current newborn screening available in Malaysia includes only G6PD deficiency, congenital hypothyroidism, and hearing loss.

Equally concerning is the lack of harmonised planning across key ministries – including MOH, the Ministry of Women, Family and Community Development (KPWK), the Ministry of Education (MOE), the Ministry of Higher Education (MOHE), Ministry of Finance (MOF), Ministry of Home Affairs, Ministry of Youth and Sports, and the Ministry of Human Resources, including their implementing agencies (such as Department of Social Welfare or Jabatan Kebajikan Malaysia (JKM), National Population and Family Development Board (or Lembaga Penduduk dan Pembangunan Keluarga Negara, LPPKN, higher learning institutions, etc). Fragmented governance

results in a disjointed continuum of care, hindering RD patients' access to inclusive education, social services, and employment opportunities.

### **Importance and Impact of the Rare Disease in Malaysia**

Delayed diagnosis can lead to irreversible physical complications and missed therapeutic windows, compromising long-term health outcomes. Insufficient clinical expertise may result in misdiagnosis, inappropriate treatment, and psychological distress for families. The financial burden of RD including but not limited to, medical expenses, frequent hospitalisation, and loss of income due to caregiving, further exacerbates household vulnerability. A fragmented, non-integrated system undermines continuity of care and leaves patients navigating multiple uncoordinated services. Moreover, broader societal and economic consequences arise when caregivers are forced to leave the workforce or when untreated RD leads to lifelong disability. If unresolved, these will widen health inequities, increase long-term healthcare costs, and dampen Malaysia's progress toward inclusive health policies that reflect the needs of all communities. This will also negatively affect national productivity and economic growth by increasing losses of disability-related workforce and long-term dependency, ultimately placing additional strain on Malaysia's gross domestic product (GDP). International estimates indicate that the exclusion of persons with disabilities from the labour market may reduce national economic output by several percentage points of GDP (approximately 3 – 7%),

through a combination of lost productivity, reduced labour participation, and increased long-term dependency (ILO 2009).

### **Intervention Approaches and Methods of Care**

Effective management of rare diseases typically involves a combination of interventions tailored to the patient's specific condition. These include early diagnosis, specialised therapies, targeted medicines (orphan drugs), genetic counselling, and long-term monitoring delivered through multi-disciplinary teams.

### **Genetic Clinics and Diagnostic Services in Malaysia**

Malaysia's genetic healthcare services span several ministries, including MOH, MOHE, Ministry of Defence Malaysia (MINDEF), and Ministry of Home Affairs (MHA). Recognising the need for research into RD, the first dedicated Genetic Medicine unit in Malaysia was established in UMMC in 1995, and scored many "firsts": first to treat lysosomal diseases (Gaucher in 1994, Pompe disease in 2007 and mucopolysaccharidosis type VI) with enzyme replacement therapy; first to offer genetic counselling and to train genetic counsellors to enable board-certification in 2003; first to establish a Malaysia Rare Disorders Society (MRDS); first public hospital to start newborn screening programme for RDs in 2015 and to offer next generation sequencing for undiagnosed malformation syndromes. Extensive research and collaborations with national and international institutions were conducted on RDs which many were hitherto unknown in the Malaysian population.

Over 120 academic journal publications were published on RDs and hereditary conditions in Malaysia, as well as pamphlets on various rare conditions. Three books on RD were published, including a compilation on patients' narratives entitled "Rare Journeys of Love",<sup>8</sup> numerous book chapters on various RDs including the Oxford Monograph in Medical Genetics<sup>9</sup> and The Institute for Democracy and Economic Affairs (IDEAS) White Paper policy document entitled "Rare Diseases in Malaysia".<sup>10</sup> Recent progress includes clinical drug trials for genetic therapeutics and policy development for RDs in Malaysia. Gene therapy was successfully used in Southeast Asia for the first time in 2020 for six UMMC patients with spinal muscular atrophy type 1, a rare genetic neuromuscular condition. Due to the efforts of first few clinical geneticists, Clinical Genetics was accepted as a paediatric subspeciality in 2006 in the National Specialist Registry. In addition to the MRDS set up as a lay support group, the Genetic Counselling Society of Malaysia, as well as Medical Genetics Society of Malaysia were established for the various professionals and genetic healthcare providers.

Clinical Genetics Service at the MOH began later in 1999 when the Genetic & Metabolic Unit was established at the Institute of Paediatrics, Hospital Kuala Lumpur (IPHKL) as one of the subspecialties in paediatrics. Since then, more paediatricians have been trained and have qualified as clinical geneticists. The Blueprint for Genetic Service was prepared by the MOH's Medical Development Division in 2008 following a series of discussions with clinical geneticists and relevant stakeholders.

The policy paper titled "Restructuring of Genetic Services in Hospital Kuala Lumpur (HKL)" was presented and accepted at the Special Meeting by the Director-General of Health on 16 December 2008. The Department of Clinical Genetics of HKL was officially established on 1 September 2009 and in Hospital Pulau Pinang (HPP) in 2019. In addition to the two MOH's Clinical Genetic Departments, a few teaching hospitals including Tunku Ampuan Besar Tuanku Aishah Rohani (Hospital Pakar Kanak-Kanak Universiti Kebangsaan Malaysia, HUKM) and Hospital Universiti Sains Malaysia (HUSM), Kubang Kerian, also offer clinical genetic services.

As of July 2025, genetic clinics operate in six major hospitals, led by an estimated 15 to 18 geneticists nationwide. The Institute of Paediatrics, HKL, functions as the national referral centre. Other key institutions include HPP, UMMC, HUKM, HUSM, and the Universiti Sultan Zainal Abidin Teaching Hospital. Several other specialist hospitals also play significant roles in RD management.

Diagnostic capacity remains limited. While the Institute for Medical Research (IMR) and Hospital Tunku Azizah of MOH offer specialised diagnostic services, including Whole Exome Sequencing and biochemical and genetic testing for inborn errors of metabolism, genetic and genomic tests are not routinely available in teaching hospitals and other parts of the country, requiring samples to be sent overseas. This creates a significant financial barrier, particularly for the B40 group, as costs are often borne by families. Although hospitals, NGOs, and

welfare units may offer partial assistance, coverage is inconsistent and insufficient, leaving many patients undiagnosed. For families who cannot afford genetic testing, the lack of diagnosis delays or prevents access to targeted treatment and clinical management. In 2015, UMMC started the first expanded newborn screening for inherited metabolic diseases in a public hospital in Malaysia, but such initiatives remain rare and have not been scaled nationally.

### **Supportive and Medical Interventions**

Supportive therapies such as physiotherapy, occupational therapy, and speech therapy are widely available but may lack sufficient frequency due to resource constraints. Community-Based Rehabilitation centres supplement hospital services by fostering social integration and enhancing functional independence.

Access to orphan drugs remains limited due to high costs and restricted financing despite the existence of MOH's technical review mechanisms and the publication of a national Rare Disease List. Even for the majority of RDs without approved orphan drugs, long-term care remains intensive and costly. Patients may require corrective surgeries, growth hormone therapy, bone marrow transplantation, specialised medical foods, assistive devices, and regular specialist follow-ups. The MOH support treatment of RDs to the tune of RM25 million in 2024.

### **Genetic and Psychosocial Counselling**

Genetic counselling supports families in understanding inheritance patterns, assessing risks, interpreting diagnostic options, and navigating long-term planning. Psychosocial counselling is equally critical due to the emotional strain of chronic caregiving, frequent hospitalisation, financial pressure, and concerns about long-term care, including "Who will care for my child when I am no longer here?". Counselling services help build resilience, address stigma, and support mental well-being across the life course.

To build local expertise in supporting patients with genetic conditions, the first core group of genetic counsellors were trained in UMMC in conjunction with the Human Genetics Society of Australasia in 2003. In 2016, UKM started the Master of Science (Genetic Counselling) programme. However, graduates face significant constraints because the MOH does not currently recognise "Genetic Counsellor" as an official position within the public healthcare system. As a result, many graduates work in research, academia, or private healthcare settings rather than in government hospitals, limiting their direct impact on clinical services for RD patients. Despite these challenges, genetic counsellors play an important role in improving awareness, providing pre- and post-test counselling, and guiding families through complex decisions related to genetic testing and disease management. Their expertise helps bridge communication gaps between clinicians and patients, supports informed consent for genomic testing, and advocates for better integration of genetic services into Malaysia's healthcare system.

### **Role of NGOs and Support Groups**

NGO-led support groups were started by parents in the mid-2000 and since then, there are currently 21 registered NGOs dedicated to issues related to RD. They provide vital emotional, informational, and advocacy support systems. They connect families, disseminate care practices, facilitate access to resources, and advocate for inclusive policies in education, healthcare, and employment. These groups help reduce isolation and strengthen community mobilisation.

### **Regular Monitoring and Research**

Continuous health monitoring ensures timely adjustments to treatment and early detection of complications. Research on RDs, including clinical trials for gene therapy and biologics, is critical but remains underfunded due to a prevailing perception that RD lacks commercial value.

Drawing on a systematic literature network analysis of RD research in Malaysia (manuscript in draft), the national research and development (R&D) landscape appears unevenly developed, with a strong historical concentration around a small number of conditions – most notably thalassaemia – where sustained research has translated into screening programmes, clearer care pathways, and population-level impact. It is noteworthy that thalassaemia is included in the Malaysian Rare Disease List 2023 due to the relatively high birth prevalence of 1 in 2000, highlighting that the research literature has been shaped by legacy public health and genetic priorities and does not, on its own, fully capture the contemporary RD

profile in Malaysia. In contrast, research on many conditions that meet the RD definition, such as metabolic, neuromuscular, and multisystem genetic disorders, remains fragmented, low-density, and largely descriptive, with limited integration across diagnostics, therapeutics, and health system domains. Emerging themes such as enzyme replacement therapies and broader “rare disease” discourse are visible but still weakly connected to national data infrastructure, implementation research, or financing mechanisms. Consequently, translation of R&D outputs to bedside care has occurred selectively and disease-specifically, rather than systemically, reflecting gaps in registries, genomic service integration, trial readiness, and RD – specific health technology assessment.

### **Institutional Consolidation and the Deepening of Expertise: Evolution**

Malaysia's institutional landscape for RDs remains relatively young, but progress is evident. As tertiary centres formalised their RD units, institutional governance and clinical protocols matured. Several hospitals established dedicated metabolic laboratories, newborn screening programmes, and molecular diagnostic units, though coverage remains inconsistent across states. Academic institutions have played a central role in advancing RD education. Universities have expanded postgraduate training in genetics, metabolic medicine, and child neurology. Despite ongoing workforce shortages, the number of specialists is gradually increasing, contributing to more balanced distribution of expertise nationwide.

Parallel to institutional growth, patient and caregiver knowledge has expanded. Civil society organisations such as MRDS and various condition-specific associations have undertaken large-scale awareness campaigns, educational workshops, and policy dialogues. Their advocacy has improved public understanding, normalised RD discourse, and reduced stigma surrounding genetic conditions, as well as advocated for non-health issues such as access to employment with dignity, child rights and protection and inclusive education.<sup>6</sup>

Although formal registries remain limited, clinicians have accumulated rich tacit knowledge from years of treating rare conditions. This tacit expertise, while insufficient as a standalone evidence base, has been instrumental in shaping clinical practice guidelines and informing policy discussions.

### **Policy Development and Governance Dynamics**

Malaysia's policy environment for RDs is characterised by incrementalism, pilot initiatives, and gradual institutionalisation rather than sweeping legislative reform. Early progress emerged through the MOH's development of the National Strategic Plan for Rare Diseases (NSPRD), which identified priorities in diagnostics, training, research, and patient care. However, implementation has faced obstacles due to limited dedicated funding, governance fragmentation, and the absence of binding statutory mandates.

### **Governance Structures**

The MOH, MOHE and academic hospitals share overlapping responsibilities for RD services. While this multi-agency system can amplify resource distribution, it often results in bureaucratic delays and inconsistencies across states. Public-private collaboration is emerging, particularly in diagnostics and enzyme replacement therapies, but remains largely ad hoc.

One notable development is the increased use of working groups comprising clinicians, MOH officers, patient representatives, and pharmacists, to standardise clinical pathways and advise on high-cost treatments. These committees have begun developing criteria for funding prioritisation, although transparency and coverage remain limited.

### **Financing Mechanisms**

The financing of RD treatments remains the most challenging policy barrier. Treatment can be broadly divided into two groups. The first group involve specific genetic or pharmacological therapeutics. Currently, it is estimated that only about 5% of RDs have a specific pharmacological agents approved. Due to the high cost of R&D of these therapeutics and the relatively small number of patients, the treatment costs are high, ranging from hundreds of thousands to millions of ringgits every year for each patient. The second group consists of non-pharmacological treatment such as specific medical foods, rehabilitation services, surgical treatment and other forms of treatments. Many of these young families have limited resources and having to cope with "out-of-pocket" expenses, thus, exacerbating the financial crises.<sup>10</sup>

Current funding models rely on:

- (i) MOH discretionary funding,
- (ii) zakat and charitable funds,
- (iii) corporate social responsibility (CSR) contributions from pharmaceutical companies,
- (iv) occasional Parliament-approved allocations.

While these mechanisms provide life-saving support for some patients, they produce inequities, unpredictability, and long waiting times. A sustainable, ring-fenced national funding mechanism has yet to be established. A number of other innovative funding mechanisms are currently under considerations and beyond the scope of this review.

### **Regulatory and Data Infrastructure**

Malaysia lacks a comprehensive RD registry, relying instead on disease-specific lists curated by clinicians. There is also no formalised national newborn screening policy beyond congenital hypothyroidism, hearing loss and G6PD deficiency. Diagnostic delays remain common, partly due to limited genomic sequencing access outside major centres.<sup>9</sup>

Nonetheless, incremental progress is visible: digital health systems under development may enable the integration of RD modules, and genetic services typically initiated their hospital-based local patient databases to monitor patients' diagnosis and treatment outcomes.

### **Synergy Between Clinical Networks and Policy Reform**

Despite structural constraints, Malaysia's RD progress

is driven by increasing synergy between clinical, institutional, and policy ecosystems. Clinical networks generate evidence and guidelines, patient groups amplify needs and lobby for resources, and policymakers respond through targeted reforms. These interactions produce: (i) improved diagnostic pathways – as Multi Disciplinary Teams (MDTs) streamline referrals and shorten diagnostic odysseys; (ii) enhanced access to therapies – as committees evaluate high-cost drugs based on accumulated clinical evidence; (iii) greater public legitimacy – as patient groups contribute to policymaking dialogues; (iv) policy experimentation – including pilot funding models, state-level newborn screening expansions, and pharmaceutical access partnerships.

Although progress is uneven, this ecosystemic synergy reflects Malaysia's shift from fragmented service provision toward coordinated governance.

### **Malaysia in the Asia-Pacific Context**

Comparatively, Malaysia is positioned in the intermediate tier of Asia-Pacific RD development. Countries such as Japan<sup>11</sup>, Taiwan<sup>12</sup>, Australia<sup>13</sup>, and South Korea<sup>14</sup> have enacted comprehensive RD laws, national insurance coverage, and advanced registries, giving them structural advantages. Meanwhile, lower-middle-income countries in Southeast Asia often lack formal RD frameworks.

Malaysia's strengths include: (i) well-developed tertiary centres with subspecialty expertise; (ii) growing advocacy infrastructure; (iii) increasing policy engagement; and (iv) regional partnerships that enhance clinician training.

However, major gaps remain in financing, statutory protections, disability classification, and genomic research integration. Addressing these gaps is essential for Malaysia to converge with regional leaders.

#### **Future Directions and Strategic Recommendations**

Malaysia's recent policy trajectory reflects an unprecedented alignment with global momentum on RDs. In May 2025, the World Health Assembly adopted a landmark Resolution on Rare Diseases, formally recognising rare conditions as a global public health priority and urging member states to strengthen national strategies, coordinated research, and cross-border cooperation. This resolution provides an international mandate for countries – especially those in the Global South – to embed RDs within universal health coverage, research systems, and long-term health financing reforms.

Building on this global shift, Malaysia launched its National Policy for Rare Diseases (2025), the country's first comprehensive framework to articulate national commitments across early detection, diagnostics, genomic integration, workforce development, data governance, and access to therapies. The policy offers a unifying national direction for what was previously a fragmented landscape of small-scale initiatives. It also establishes the governance mechanisms needed to integrate RDs into broader health planning, including research and innovation agendas.

This momentum is further reflected in Malaysia's health research ecosystem. For the first time, RDs have been included in the deliberations for the 13<sup>th</sup> Malaysia Plan Health Research Priorities (2026–2030). While the formal document has not yet been released, the stakeholder engagement process led by the MOH has emphasised that RDs form part of the country's emerging research concerns. The consultative technical discussions spanning clinicians, researchers, policymakers, and patient groups, signal a significant normative shift: RDs are no longer viewed as marginal, but increasingly recognised as a legitimate component of national research planning. Any reference to RDs in the forthcoming priority list should, however, be interpreted cautiously until the official document is published.

At the regional level, Malaysia has signalled its intention to play a leadership role in advancing RD policy within Southeast Asia. During the Southeast Asia Rare Disease (SEA-RD) Policy Forum, Malaysia publicly committed to spearheading the development of an ASEAN Rare Disease Declaration, positioning RDs as a shared regional health and equity concern rather than a series of isolated national challenges. This commitment reflects an emerging recognition that coordinated policy frameworks, cross-border data sharing, and collective advocacy are essential to addressing the structural disadvantages faced by people living with RDs across the ASEAN region (The Star 2025).

To sustain progress, Malaysia's RD ecosystem requires structural reforms across five domains which were outlined in the NPRD:

1. **Legislative and Policy Consolidation:** A dedicated Rare Disease Act, as practised in Japan and Taiwan, would strengthen rights-based access, financing continuity, and multi-agency accountability.
2. **Comprehensive National Funding Mechanism:** A ring-fenced RD fund, financed through blended models (government budget, social insurance, public-private partnerships), is critical for equitable access to high-cost therapies.
3. **National Rare Disease Registry and Genomic Integration:** A unified registry supported by genomic sequencing platforms would generate high-quality epidemiological data and inform precision-medicine-based policy decisions.
4. **Expansion of Newborn Screening:** Scaling newborn screening beyond the existing limited panel would reduce long-term morbidity, lower economic burden, and improve survival for treatable metabolic disorders.
5. **Strengthening the Workforce Pipeline:** Developing specialised training tracks, fellowships, and regional rotations would expand the talent pool in genetics, metabolic medicine, dietetics, and genetic counselling.

## Conclusion

Malaysia's ecosystem of RD care reflects both significant progress and persistent structural challenges. While diagnostic, clinical, and community-based resources exist, systemic gaps particularly in financing, inter-ministerial coordination, public awareness, and research investment, remain a hindrance to equitable access to care. Strengthening nationwide newborn screening, expanding genetic services, recognising genetic counselling, increasing funding for orphan drugs, and establishing a national RD registry are essential steps toward a more inclusive and responsive healthcare system. Ultimately, reframing the RD community not as passive beneficiaries of welfare but as citizens entitled to equal rights and human dignity is fundamental for building a society where no individual is left behind.

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