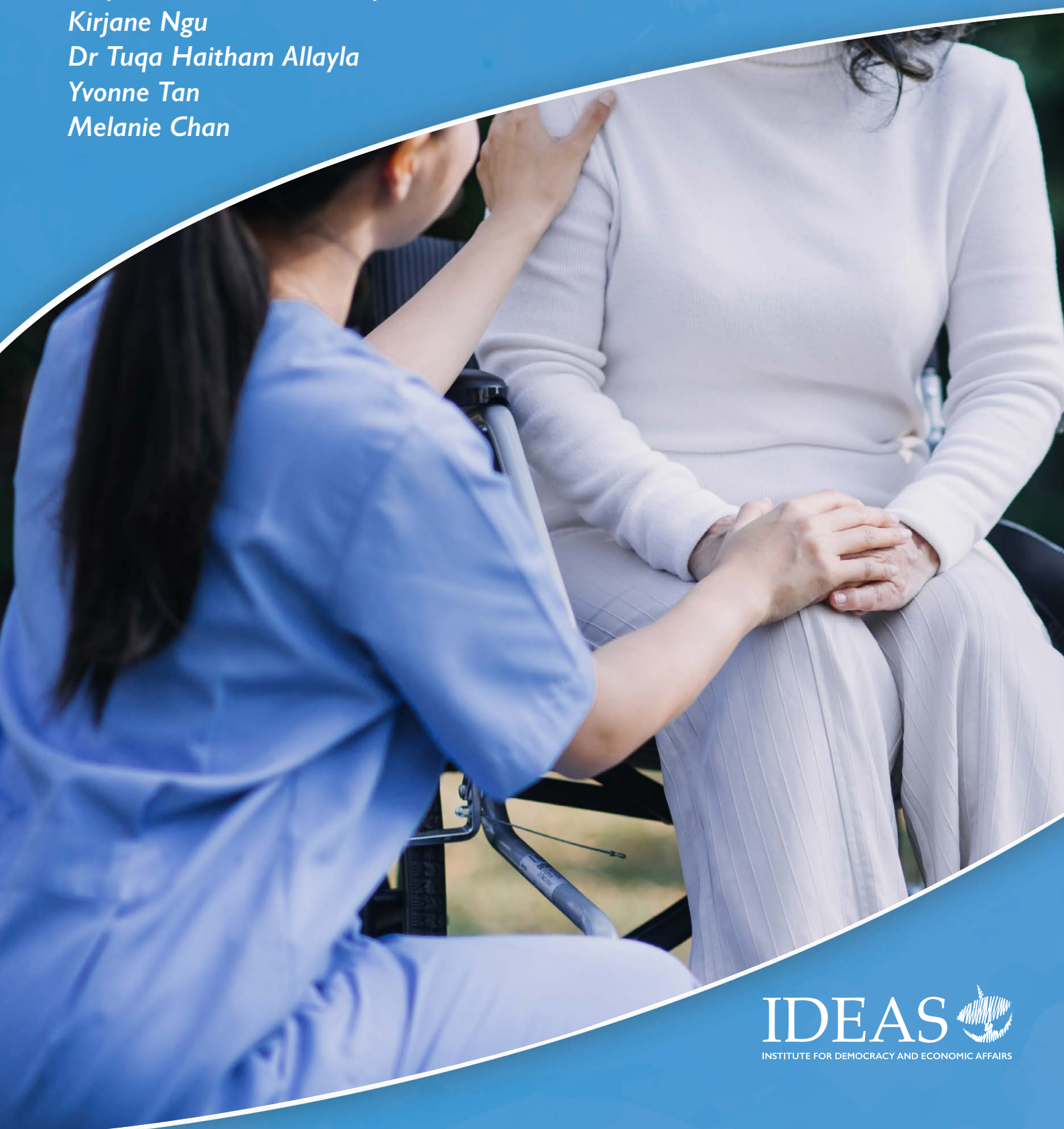
Policy Ideas
№ 78
October 2023

Solidarity Amidst Rarity: Reforms for Equitable and Sustainable Rare Disease Management

Professor Asrul Akmal Shafie
Kirjane Ngu
Dr Tuqa Haitham Allayla
Yvonne Tan
Melanie Chan



IDEAS
INSTITUTE FOR DEMOCRACY AND ECONOMIC AFFAIRS

Authors

Professor Dr. Asrul Akmal Shafie is a Professor of Pharmacoeconomics (first in Malaysia) at the School of Pharmaceutical Sciences, and the Director of the Institutional Planning & Strategic Centre, Universiti Sains Malaysia (USM). He is the Co-Editor of Value in Health Asian Region, and a member of the editorial board for Pharmacoeconomics journal. He is the recipient of the prestigious International Fellowship for International Society of Pharmacoeconomics and Outcomes Research, Young Scientists for the Academy of Sciences Malaysia, Cancer Research Award by the Cancer Council Malaysia, and Top 2% in the World in their Respective Field by Stanford University Higher Education Global Impact Ranking.

Kirjane Ngu is a Senior Research Executive at the Social Policy Unit of IDEAS Malaysia. Her research and advocacy focus on cross-cutting social policy and development areas such as health, social protection, employment, education, and human rights, particularly for vulnerable groups such as children, youth, persons with disabilities, indigenous groups, refugees and asylum-seekers, in both the national and regional context such as ASEAN. She has published various research papers on health systems, community health, youth development and clinical neuropsychology. She holds a Bachelor of Science degree in Psychology from the Institute of Psychology, Psychiatry, and Neuroscience, King's College London.

Dr. Tuqa Haitham Allayla is a dedicated researcher with a Ph.D. in Pharmacy Practice from the School of Pharmaceutical Sciences at Universiti Sains Malaysia (USM). Her research revolves around Pharmacy Practice, with a focus on Extended Pharmacy Services, Qualitative Research, Digital Health and Public Perceptions. Notably, she has achieved success in publishing multiple booklets and papers. She actively contributes to academic discourse as a reviewer for various journals and conferences. Her commitment to advancing pharmacy practice through innovative research underscores her valuable contributions to the field.

Yvonne Tan is a Senior Research Executive at the Social Policy Unit of IDEAS Malaysia. Her research interests include labour and migration, health and socioeconomic equity, and political economy within the Southeast Asian region. Yvonne's experience spans health systems, public and community health from her engagements with United Nations University-International Institute for Global Health, Research Triangle Institute, Health Equity Initiatives and Monash University Malaysia. She also believes our past informs our future and works interdisciplinarily having published on cultural studies related to dynamics of challenging imperial inequalities and processes of political change. Her insights have been featured on platforms including Malaysiakini and MalaysiaNow. She graduated with a Masters in Southeast Asian Studies from Goethe University Frankfurt.

Melanie Chan is a Research Executive under the Social Policy Unit at IDEAS Malaysia. She achieved the accolade of the Best Graduate Award for her Bachelor of Arts and Social Sciences degree from Monash University Malaysia, where she pursued a dual major in Global Studies and Communications. Having undergone a prior internship at IDEAS, she has since realised that her true aspiration lies in harnessing the power of the written word to inspire and hopefully, realise inclusivity across all facets of society.

Acknowledgements

This report is part of a project on Rare Disease Management in Malaysia, managed by Kirjane Ngu. The authors would like to thank Dr. Juita Mohamad for supervising this project, and Dr. Tricia Yeoh for her guidance and her support in reviewing the report. We would also like to thank Faiz Zaidi for his support in project management, Zuhdi Farhan in event management and communications, and Teck Hee for the design of this publication. We would also like to thank our interns, particularly Laura Zhang, who have assisted in the qualitative analysis for this report.

We are indebted to the survivors, caregivers and patient-advocacy groups for graciously sharing their stories, experiences and challenges in the focus group discussions and interviews. We are also deeply grateful for the contributions and support of representatives from the following organisations:

- Rare Disease Alliance Foundation Malaysia
- Malaysian Rare Disease Disorder Society
- Persatuan SLE Malaysia, Persatuan Pesakit Motor Neuron Malaysia
- DEBRA Malaysia
- Persatuan Spinal Muscular Atrophy Malaysia
- Pertubuhan Penyakit Lisosomal Malaysia
- Persatuan Sindrom Prader-Willi Malaysia
- Kuala Lumpur and Selangor Albinism Association
- Pertubuhan Kebajikan Pesakit Inchthyosis Malaysia
- Persatuan Multiple Sklerosis Malaysia
- Malaysian Patient Organisation for Primary Immunodeficiencies
- Hospital Kuala Lumpur
- Universiti Kebangsaan Malaysia
- Universiti Malaya
- Bahagian Perkembangan Perubatan, Kementerian Kesihatan Malaysia
- Bahagian Amalan dan Perkembangan Farmasi, Kementerian Kesihatan Malaysia
- Bahagian Dasar dan Perancangan Strategik, Kementerian Pembangunan Wanita, Keluarga dan Masyarakat

We would also like to thank Professor Dr. Thong Meow-Kong and Alissa Marianne Rode for reviewing this report.

This project was supported by AstraZeneca Sdn. Bhd. and commissioned by IDEAS. However, the opinions expressed in this report are those of the author(s) and do not necessarily represent IDEAS or AstraZeneca Sdn. Bhd.

Table of Contents

Executive Summary

While the accessibility of healthcare in Malaysia has undoubtedly progressed considerably in recent years, the same cannot be said for patients grappling with rare diseases. Due to a combination of a lack of public awareness, a subpar standard of knowledge among professionals, and a dearth of diagnostic facilities, rare disease patients face significant delays in healthcare access. With the majority of healthcare facilities and expertise concentrated in the Klang Valley, location has become a prohibitive physical barrier for those residing outside the vicinity.

Furthermore, the high costs of diagnostic tests and treatment for rare diseases are not routinely funded by the government, necessitating short-term funding from external schemes, or requiring out-of-pocket funds which potentially leaves rare disease patients economically vulnerable. This coupled with the lack of entitlement to the usual forms of social healthcare protection, result in rare disease patients being left to fend for themselves at the expense of their physical, financial, and emotional well-being.

In order to identify and measure healthcare access for rare diseases throughout the patient pathway, this study employed Levesque's framework to conceptualise healthcare access from the perspectives of the healthcare system and patients. Based on these barriers, this paper recommends four critical pillars to improve healthcare access for rare disease patients and stratifies them based on their complexities. The contents and the policy recommendations put forth in this report are primarily based on findings from two focus group discussions with patients, and one roundtable discussion with stakeholders. Hence, the recommendations were influenced by their input, which inadvertently skews towards a patient-centric view.

In the short-term, Malaysia should enhance its healthcare resource capacity (Pillar 1) through collaborative capacity building, resource sharing and the creation of a patient navigation platform to link healthcare facilities with various stakeholders.

In the mid-term, Malaysia should seek to create a comprehensive and sustainable framework for rare disease financing (Pillar 2) through legislated incentives and negotiated cost containment. As many of the solutions require interministerial collaboration, the governance system should be strengthened in order to achieve a holistic management of rare diseases (Pillar 3).

Lastly, in the long-term, the country's social protection systems (Pillar 4) need to be enhanced to empower and facilitate the resilience of individuals with rare diseases along with their caregivers. The implementation of these initiatives requires strong commitment, political will, and a driven champion to bring the necessary changes to fruition.

Strategic Pillars	Aspects of Policy Development	Timeframe
 **Pillar 1: Enhancing healthcare resources capacity**	Building the capability of healthcare professionals through collaboration, strengthening the role of universities and prioritising government position warrants	Short-term *(1-2 years)*
	Resource sharing for diagnostic tests between interministerial facilities	
	Improving seamless collaboration and integration between healthcare facilities and patient groups	
 **Pillar 2: Comprehensive and sustainable rare disease financing**	Cultivating the orphan drugs market through regulatory incentives	Mid-term *(3-4 years)*
	Ensuring affordability of treatment through risk sharing mechanisms	
	Leveraging pooled procurement to boost purchasing power	
	Accelerating the Health Technological Assessment (HTA) process through joint assessment	
	Strengthening the role of the trust fund to address unmet needs of rare disease treatment affordability	
	Developing the pharmaceutical industry to promote generic competition for drugs treating rare diseases	
 **Pillar 3: Strengthening governance for holistic management of rare diseases**	Institutionalising a legal framework for rare diseases to protect patients from heightened vulnerability	
	Interministerial coordination and collaboration as key to improving integration and continuity of care	
 **Pillar 4: Enhancing social protection systems to promote resilience of individuals with rare diseases and their caregivers**	Protecting rare disease patients and householders with improved insurance coverage	Long-term *(more than 5 years)*
	Improved coverage and assistance for support services and assistive devices	
	Promoting inclusive employment opportunities for people living with rare diseases and their caregivers	

Background

Importance of Equitable Access to Quality Healthcare for Rare Diseases

Access to quality healthcare can be defined as the opportunity to identify, seek, reach, obtain, or use healthcare and to ensure that the needs for these services are fulfilled. It encompasses the adequacy of the various aspects of the healthcare system, and also any consequences it may pose to patients.

Equitable access to quality healthcare embodies the holistic values of care and compassion in the Malaysia Madani framework. It also supports the United Nations resolution that recognises 'the need to promote and protect the human rights of all persons, including the estimated 300 million Persons Living with a Rare Disease worldwide'.[1]

The impact of accessible and quality healthcare on patients, the health system, and society as a whole is substantial. The healthcare system will benefit from reduced morbidity and mortality, while patients will have more promising outcomes, which translates into better quality of life. In turn, society will benefit from improved productivity and lower healthcare costs.

Yet, for rare disease patients, access to quality healthcare remains a distant dream. A rare disease, also known as an orphan disease, is a health condition that affects a small portion of the population. Globally, it is estimated that between 350 and 475 million people live with a rare disease,[2] with approximately 5,000 to 8,000 types of rare diseases in existence.[3] These conditions can be caused by genetic mutations, environmental factors, or a combination of both. Many rare diseases have limited treatment options, and they often go undiagnosed or misdiagnosed for years. It was estimated that less than 10% of existing rare diseases have an effective treatment. As most countries can only estimate the prevalence of rare diseases, the lack of accurate epidemiological data has hindered the management and treatment of such.

This report focuses on the challenges faced by patients across the rare disease treatment pathway building on IDEAS' previous work on rare disease policy. They include *Policy Paper No 67—Next Steps for Rare Diseases in Malaysia: Improving Access to Treatments* (2020)[4], *Whitepaper: Rare Diseases in Malaysia* (2019)[5], *Policy IDEAS No 58—Improving Access to Orphan Drugs in Malaysia* (2019)[6] and *Brief IDEAS No 10—National Policy on Rare Diseases Living with Dignity: In Search of Solutions for Rare Diseases* (2018).[7]

Table 1: Number of rare disease patients in Malaysia (based on 10 major tertiary hospitals in Malaysia).

Disease/Group of conditions	Numbers in Malaysia
Systemic lupus erythematosus (SLE)	43 patients[8]
Spinal muscular atrophy (SMA)	9 patients[9]
Osteogenesis imperfecta (OI)	45 patients[9]
Maple Syrup Urine Disease (MSUD)	33 patients[9]
Citrullinemia type II (citrin deficiency)	33 patients[9]
Duchenne Muscular Dystrophy (DMD)	26 patients[9]
Marfan syndrome	81 patients[9]
Prader-Willi syndrome	60 patients[9]
Noonan syndrome	16 patients[9]
Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes (MELAS)	39 patients[9]
Leigh syndrome caused by mutations of nuclear genes	32 patients[9]
Myoclonic epilepsy associated with ragged-red fibres (MERRF)	12 patients[9]
Hunter syndrome or Mucopolysaccharidosis type II (MPS II)	39 patients[9]
Pompe disease or Glycogen storage disease type II (GSD II)	17 patients[9]
Multiple Sclerosis	8 patients[9]
Cystic Fibrosis	9 patients[9]
Haemophilia A	24 patients[9]
Primary Pulmonary Arterial Hypertension	1 patients[9]
Fabry Disease	2 patients[9]

The burden of rare diseases can be far-reaching and have a significant impact on both individuals and society. For those affected by a rare disease, the physical, emotional, and financial toll can be overwhelming. Physical symptoms can be debilitating and cause significant pain and discomfort, while emotional distress can result from the isolation and stigma often associated with rare diseases. Additionally, the financial burden of managing a rare disease can be substantial, as many rare diseases have no approved treatments and require expensive and ongoing medical care.

When discussing society as a whole, the lack of investment and research in rare diseases can lead to significant challenges in diagnosing and treating these conditions. As a result, many rare diseases remain poorly understood and lack effective treatment, thus placing a burden on healthcare systems, and constitutes a drain on limited resources.

Many of the rare disease patients, often alongside their families, face discrimination that eventually leads to social isolation and exclusion. They may face ensuing problems integrating with mainstream education and the economic system, leading to the loss of learning and earning opportunities.

Research related to rare diseases is important because it can help to improve health outcomes for individuals with these conditions and improve their quality of life through effective and accessible medical care. Finally, raising awareness about rare diseases and the challenges faced by affected individuals can lead to increased understanding, support, and investment in medical research, ultimately benefiting society.

This policy paper will provide some insight into the barriers and challenges faced by rare disease patients in accessing optimal treatment across the rare disease treatment pathway. The findings outlined in this policy paper can guide policymakers, healthcare professionals, and advocacy organisations towards the appropriate allocation of resources in the healthcare system to improve patient access to treatments, while also ensuring that healthcare providers are given the necessary training and resources to diagnose and manage these conditions effectively.

Key Barriers and Challenges to Accessing Optimal Treatment

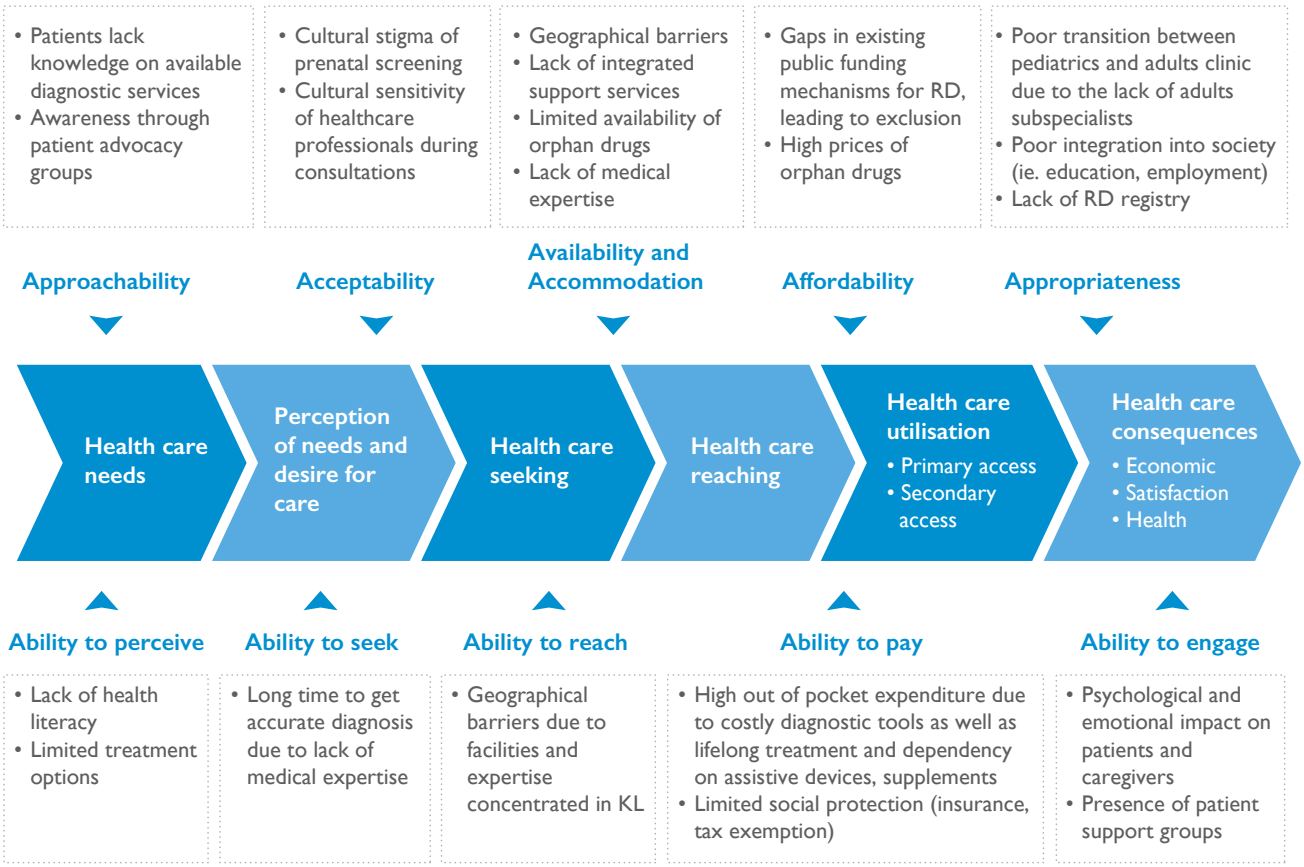


Figure 1: Access to Healthcare Conceptual Framework (adapted from Levesque et al, 2013).

In order to help identify and measure healthcare access for rare diseases, it is important to organise them into a robust theoretical framework. This study used Levesque's framework to conceptualise healthcare access for rare diseases.[10] Unlike other frameworks, Levesque's framework allows both the healthcare system and patients' perspectives to be taken into account when assessing healthcare access. This will facilitate an assessment of the difficulties of healthcare access from two perspectives: the shortcomings of the healthcare system and the patient's own capacity and awareness.

The framework categorises healthcare system factors into five dimensions, i.e. approachability, acceptability, availability and accommodation, affordability, and appropriateness. The patient factor takes into account the population's socioeconomic determinants resulting in the incorporation of the five corresponding abilities of individuals and populations: to perceive, to seek, to reach, to pay, and to engage in healthcare.

1. HEALTHCARE SEEKING

For a patient to seek healthcare for their rare disease condition, they need to be able to perceive the need for care. Perceiving the need for care, which is often the first step in the patient's healthcare journey, is determined by the patient's knowledge (encompassing aspects like basic health literacy), as well as existing beliefs (including cultural and social factors) about health and sickness. In addition to the patient factors, some features of the healthcare system could affect the process, including the healthcare providers' ability to provide information and education to patients and their families regarding the availability of relevant services and treatment.

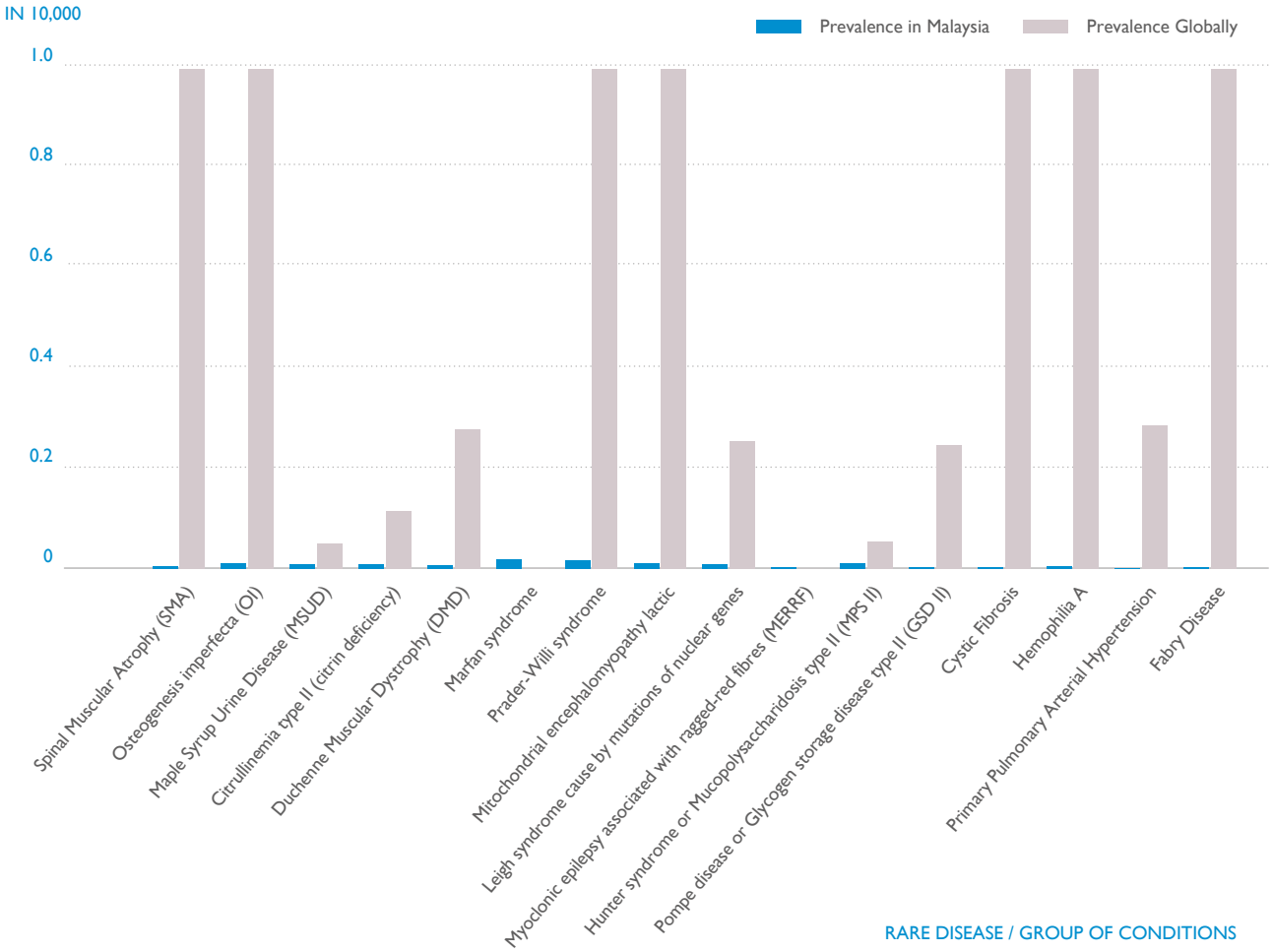


Figure 2: Comparing Prevalence in Malaysia and Prevalence Globally.

Rare disease patients frequently find themselves trapped, oblivious of where or how to begin seeking proper treatment for their condition. In addition, the average time for an accurate diagnosis of a rare disease is about 4–5 years.[11,12] The prevalence of rare diseases in Malaysia is lower than global estimates as shown in Figure 2, and this could be the result of underdiagnosis. There are a number of factors that contribute to this discrepancy.

1.1 Lack of health literacy among the public

Limited knowledge of health-related information can render it difficult for patients to recognise their conditions and communicate their needs and preferences to healthcare providers. The focus group discussions found that most patients and caregivers had not heard about rare diseases nor encountered other rare disease patients before either experiencing the symptoms themselves, or becoming rare disease caregivers.

Without a medical or educational background to assist them in expressing their opinions, patients with rare diseases often have difficulty explaining their medical condition or symptoms to healthcare providers. This leaves patients reliant upon self-education by seeking necessary health-related information either via online platforms or various support groups, in order to have productive conversations with healthcare providers.

> ***P4: "At first, I can't [sic] speak with the doctor using medical terms, but as I chat [sic] with other patients (via FB group) about the symptoms, I learned to speak to my doctor in medical terms. Like GI bleeding, IVIG, subcutaneous, intravenous."***

According to the National Health and Morbidity Survey 2019 conducted by the Ministry of Health Malaysia, 1 in 3 Malaysian adults had low health literacy.[13] Low health literacy could affect patients' perceptions of their healthcare needs, hamper their ability to navigate the healthcare system, and hinder any benefits expected from healthcare providers when discussing their treatment plan, all of which could lead to poorer health outcomes.

Similar findings were reported in various instances, such as a study in which rare disease patients were found to have a low level of illness perception due to the lack of knowledge and information about their rare disease condition.[14] Furthermore, lacking a basic understanding of the disease is often linked with a negative quality of life which may arise due to the anxiety of not having a definitive diagnosis or a treatment plan.[15] Consequently, independent health information-seeking behaviour is a common and necessary practice among rare disease patients and their caregivers due to the inadequacy of health-related information provided.

1.2 Limited knowledge of rare diseases (Healthcare Professionals)

A patient's willingness to access healthcare services is directly linked to the effort on the part of healthcare professionals to provide adequate information on the availability of related services and speciality centres. Therefore, healthcare providers with a limited understanding of rare diseases can limit the patients' perception of healthcare needs, leading to delays in obtaining the correct diagnosis.

> ***P6: "When my son was born in a government hospital, the nurses and doctors had not heard of Epidermolysis Bullosa Simplex (EBS) or seen a baby with skin being peeled [sic] off. When I approached [sic] my son's Epidermolysis Bullosa, I actually know [sic] all related information through Google."***

Many patients with rare diseases often struggle to find physicians knowledgeable about their conditions, leading to misdiagnosis, delays in the correct diagnosis, and delays in receiving appropriate treatment. A similar pattern was reported in another study whereby rare disease patients and their families noted the most common causes of diagnosis delay to be related to physicians' lack of medical knowledge.[16] It is challenging for patients to identify a suitable healthcare service, particularly if the correct diagnosis was delayed. The physician interviewed pointed out that low awareness of various rare diseases and treatment options makes it difficult for those affected to recognise their need for help.

> ***P17: "These are the patients who are already diagnosed, they know where they can go to get treatment. But there are many who still don't know their diagnosis, they will go searching for the clinic, they may not [have been to a] genetic clinic if they have not been diagnosed yet."***

Another common challenge that rare disease patients must endure is having to visit many physicians before receiving a confirmed diagnosis. Many patients and caregivers had highlighted that diagnosis and ongoing care are often provided by multiple health centres, which can be challenging to navigate, further complicating the rare disease patient's treatment journey.

> ***P1: "One thing that I learned is that we have to go to many experts, I didn't know that. One of the parents I know advise [sic] me on that. Thus, I went visiting many different doctors to ask about the risk of the treatment. I actually went to three different doctors in Malaysia, and three different doctors in the US."***

1.3 Lack of training facilities

In the Asia-Pacific region, one of the main challenges related to the rare disease patient treatment journey is the limited training of healthcare providers.[17] The lack of local training facilities in Malaysia was reported as one major reason for the lack of knowledge among medical doctors. From the roundtable discussion with experts and patient groups, it was highlighted that rare disease specialists are scarce in the country as proven by the fact that many rare disease patients are being treated by general specialists. However, there are certain rare disease groups that need to be attended by clinical and allied care sub-specialists (e.g. paediatric geneticist and genetics counsellors).

In Malaysia, there are 38 institutions that offer medically-related programmes.[18] There are seven university teaching hospitals, namely University of Malaya Medical Center (PPUM), National University of Malaysia Medical Center (PPUKM), Hospital Sultan Abdul Aziz Shah (HSAAS UPM), Sultan Ahmad Shah Medical Centre @ International Islamic University of Malaysia (SASMEC @ IIUM), Universiti Sains Malaysia (USM) Hospital, Hospital Universiti Malaysia Sabah (UMS) and UniSZA Teaching Hospital (HPUniSZA.).[19] Only in recent years has knowledge of rare diseases been integrated into the medical curriculum.

According to the Genetic Counselling Society Malaysia, there is one genetic counselling postgraduate programme in Malaysia (offered by Universiti Kebangsaan Malaysia) that is designed to prepare postgraduate students for clinical practice in the field of genetic counselling. The nation also has seven genetic services and training centres to train genetic counsellors.[20]

Besides that, according to the Ministry of Health (MOH), genetic counselling services are only provided in Kuala Lumpur Hospital (HKL) and Penang Hospital.[21] These genetic counselling sessions are conducted by nurses at genetic specialist clinics, under the close supervision of a clinical geneticist. There are no genetic counsellor service schemes established under the Public Service Department (JPA) as of the time of writing. However, the Ministry of Health has plans to review the proposed establishment of this scheme (genetic counselling) in an effort to produce more genetic counsellors under the ministry.

Moreover, limited knowledge and scarcity of resources has led to a shortage of trained healthcare professionals in the area of rare diseases. The interviewee highlighted the shortage of trained personnel in rehabilitation centres.

> ***P18: "So there are some centres, they can only cater [for] one child for three sessions a week, or two sessions per week, because there's [sic] not enough trainers. And these trainers are volunteers and most of them have only SPM level. They don't have [the] qualification to train these disabled children. So it's very sad that these children are neglected in terms of rehabilitation."***

Rehabilitation plays a vital role in improving the quality of life for patients. It has been found that patients with Systemic Lupus Erythematosus (SLE) require physical rehabilitation to manage their condition by reducing muscle weakness while simultaneously increasing muscle endurance. A study has shown that graded aerobic exercise rehabilitation programmes can enhance energy levels, cardiovascular fitness, and muscle strength in SLE patients. It is recommended that these sessions be conducted three times a week, with each session lasting 30 to 50 minutes, in order to achieve the desired outcomes.[22]

In the case of a patient with Pompe disease, a rare genetic disorder that affects the muscles—particularly the heart and skeletal muscles—the primary goal of physical rehabilitation is to maintain and improve muscle strength, enhance mobility, and address respiratory issues. The proposed rehabilitation programme for managing Pompe disease consists of a 12-week training programme with 36 sessions of aerobic, resistance, and core stability exercises. These sessions have been reported to increase muscular endurance and strength.[23]

1.4 Lack of diagnostic facilities

While a knowledgeable clinician is undoubtedly required for an accurate diagnosis, the availability of the right equipment is also fundamental. Approximately 80% of these rare diseases are estimated to be genetic in origin and 75% of rare diseases affect children.[24] Genetic testing and DNA sequencing technologies play a crucial role in detecting specific genetic variations or mutations associated with rare diseases.

Genetic testing encompasses a wide range of techniques and technologies, and the specific equipment used can vary depending on the type of testing being performed. This results in the ensuing high infrastructure cost due to the need to cover special equipment or instruments, reagents, and human resource skills and training.

In earlier years Malaysia severely lacked the capacity for in-house testing and diagnostic facilities, thus rare disease tests had to be sent to Australia, Japan, and Taiwan for analysis; this increased the total cost of treatment. However, since 2014, selected rare disease-related tests have been available at the Institute for Medical Research, which enabled a cost reduction from AUD2,500 to only AUD240–AUD750 (RM800–RM2,000).[9] Ministry of Health has also allocated RM250,000 to HKL since 2008 for the outsourcing of diagnostic tests for patients suspected of having rare diseases.[25]

It was reported that in Africa and the Middle East, one of the main challenges affecting the quality of care and treatment for rare disease patients is the lack of capacity and diagnostic resources in both public and private sectors.[26] A delay in diagnosing the condition contributes to the disease progression, which inadvertently causes deteriorating health outcomes. For instance, in Egypt, myeloproliferative neoplasms are often diagnosed in later stages due to difficulties in distinguishing this disease from more common conditions.[27]

Global evidence indicates that the diagnostic process is often a lengthy journey as rare disease patients have to endure multiple and often repetitive interactions with healthcare providers and specialists before obtaining an accurate diagnosis. Even when effective treatment is available, the delay and inaccuracy of diagnosis prevents patients from accessing the treatment and care they need.[15] This finding is exemplified by a patient who was unable to access the necessary treatment due to an incorrect diagnosis.

> ***P5: "I sent my daughter to private Hospital X in KL. But that was a mistake actually. When she got left eye blindness in the early stage I sent her to Hospital X, [there are] only about three or four specialists at the Hospital X. They diagnose [sic] her as an optic neuritis. Let's just give her steroids. The specialist didn't even refer to other doctors. Should I [have] brought her to a government hospital or a teaching hospital? Maybe her left eye could have been saved. They came up with a semi-proper diagnosis. They were still not certain until recently when they did the genetic test and she was diagnosed as ANCA-negative Vasculitis."***

Likewise, in Latin American nations, rare disease patients often endure a long and arduous journey before being granted a confirmed diagnosis and an effective treatment plan. In countries like Brazil, Colombia, and Argentina, it was reported that rare disease patients and their caregivers often grapple with various challenges when seeking ongoing care. These include a history of numerous interactions with healthcare facilities and specialties, late or incorrect diagnoses, and lack of a treatment plan.[19] Patients in Colombia have to seek a minimum of eight consultations before receiving an accurate diagnosis, receiving at least three incorrect diagnoses along the way.[28]

Similarly, in Europe, a study highlighted that patients have a misdiagnosis more than once in over 40% of cases.[29] As such, the lack of diagnostic facilities and capabilities in the country is a critical obstacle in accessing accurate and timely diagnosis.

1.5 Lack of newborn screening

Congenital abnormalities are the second leading cause of death among neonates in Malaysia.[30] An expanded newborn screening (NBS) programme is an important tool for the early diagnosis and prevention of lifetime impairment for many rare disease patients. It includes screening for haematological, metabolic, and hormonal disorders. Most screening panels include various inborn errors in the metabolism of amino acids, fatty acids, organic acids, glucose-6-phosphate dehydrogenase deficiency (G6PD deficiency), and congenital hypothyroidism.[31]

The World Health Organisation (WHO) recommends that all countries establish NBS programmes to promote the primary prevention of congenital anomalies. Unfortunately, limited local facilities are equipped with the capacity to run these tests, which necessitates a transfer of the specimen to an overseas laboratory for testing. As genetic disease can be hereditary, the test should also be extended to the patient's immediate family. At present, Malaysia has not yet implemented an expanded NBS programme.

1.6 Time taken to get an accurate diagnosis

Getting an accurate diagnosis for rare disease patients can be a long and challenging journey, as stated by most participants.

> ***P2: "It took nine years for my wife until she got the correct diagnosis."***
>
> ***P5: "It took about nine years for my daughter to get a proper diagnosis after [a] genetic [test]. For her case, simply because the name of the disease [she suffers from] was not in the list*. Then they don't know where to put her. Then they put her under SLE, but she's not SLE. She was [an] Anca-negative Vasculitis patient."***
>
> ***Note: Anca-negative Vasculitis was not listed as a rare disease. Thus, she was not eligible to get the funds for Immunotherapy Treatment (IVIG) at first.***

Even in developed regions with more comprehensive healthcare systems, such as North America, rare disease patients often experience lengthy delays before receiving a diagnosis, requiring multiple interactions with the healthcare system. For example, in Canada, 20% of rare disease patients wait between 6 to 14 years to get diagnosed, with 60% of them consulting 3 to over 20 specialists before receiving a correct diagnosis.[32] In the United States, rare disease patients need an average of 17 healthcare interactions to obtain a proper diagnosis,[33] while in Europe, 1 in 4 rare disease patients may wait a period of 5 to 30 years to get a diagnosis. [28]

Despite medical advances and technological innovations, delayed diagnosis remains a persistent challenge for rare disease patients worldwide, primarily due to the limited access to specialised testing and expertise, alongside the complexity of rare diseases.

2. HEALTHCARE REACHING

Once a patient has learned of their condition, reaching the appropriate healthcare facility—one which offers specialised care—could be challenging. A patient's capacity to reach healthcare is related to the notion of personal mobility and transportation, in addition to the availability and accommodation of healthcare providers or facilities. A patient should be able to physically reach healthcare without undue delay. Unfortunately, many of the specialised care facilities, especially within the Ministry of Health, are concentrated in the Klang Valley.

The Genetic Department of Hospital Kuala Lumpur, which was established in 2009, currently has six paediatric clinical genetic specialists. It was previously the only specialised service available in Malaysia up until 2019, when the genetic service was established in Hospital Pulau Pinang. With two paediatric clinical genetic specialists, Hospital Pulau Pinang now serves as a referral centre for the northern region.

> ***P16: "We should be getting services to patients wherever they are, and this is a big problem because we are not reaching out to all patients. There is no equity in the sense of healthcare for this group of patients. If you are born with a rare condition in Terengganu you're in a bit of trouble because you might not get diagnosed until five years later, and then you have to travel either you go to HUSM or drive all the way to HKL. So it's actually not in the strategic interest of the country."***

This constitutes a major inconvenience for patients who often require lifetime treatment. For example, a paediatric patient on an Enzyme Replacement Therapy (ERT) requires Intravenous (IV) Infusion of a solution that contains an enzyme that is deficient or absent in the body. Since ERT treatment is a lifelong treatment, it should be consistently administered throughout the patient's life once a patient has begun the treatment, which inevitably requires the patient to make multiple visits to the healthcare facility. Another example is a patient with ANCA-negative Vasculitis who requires weekly follow-up visits to manage their rare disease condition through Immunotherapy Treatment (IVIG).

Moreover, even when treatment is available, it is usually not geographically accessible, with many rare disease patients being forced to travel long distances (sometimes outside their region) to access treatment, which incurs additional costs. This is particularly problematic when rare disease patients, alongside their caregivers, often find themselves absent from work to attend treatment sessions.

> ***P6: "In Sabah or Sarawak, patients are more like "we take care of ourselves", because they can't approach the hospital because it's too far away. It might take them about half a day to make a hospital visit without getting any good result or ideas on how to deal with their health condition."***
>
> ***P4: "I think most of the rare disease patients, usually they just give up the health service because of the location. It's hard for them because they had [sic] to wait. Sometimes [for] the parents also because of time and work. So, that's a limitation."***

Global evidence also supports these findings. In Europe, for example, many rare disease patients encounter obstacles in treatment access and availability, with nearly a quarter unable to access treatment in their own country because treatment is simply unavailable.[34] Additionally, 1 in 4 rare disease patients must travel to a different region to be diagnosed.[35] In Argentina, even with proper diagnosis, rare disease patients are forced to travel to different hospitals to undergo specific tests. The main obstacles here include transportation difficulties and long distances due to the concentration of most healthcare facilities in large urban areas.[15]

In the Asia-Pacific region, similar challenges exist, including a scarcity of facilities and clinical expertise for rare diseases, with only a few health centres offering specialised services. For example, a 2016 study in Thailand reported that most of the geneticists were located in major cities,[17] while another study found that 43% of the orphan drugs approved in the United States did not receive approval in Japan.[36] Consequently, rare disease patients often find themselves compelled to travel both domestically and internationally in pursuit of the necessary treatment.

3. HEALTHCARE UTILISATION

Once their healthcare needs are determined, patients will be prescribed appropriate treatment for the management of their rare disease. There are more than 7,000 types of rare diseases, of which merely around 500 have been identified in Malaysia. Treatment for rare disease patients varies widely and could include Enzyme Replacement Therapy (ERT), gene therapy, orphan drugs, biologics, food supplements, and rehabilitation.

Estimates show that there are 12,961 rare disease patients in Malaysia. Out of this number, a survey conducted in selected government facilities predict that only 60% of rare disease patients receive their treatment (see Table 2) in Malaysia.[5]

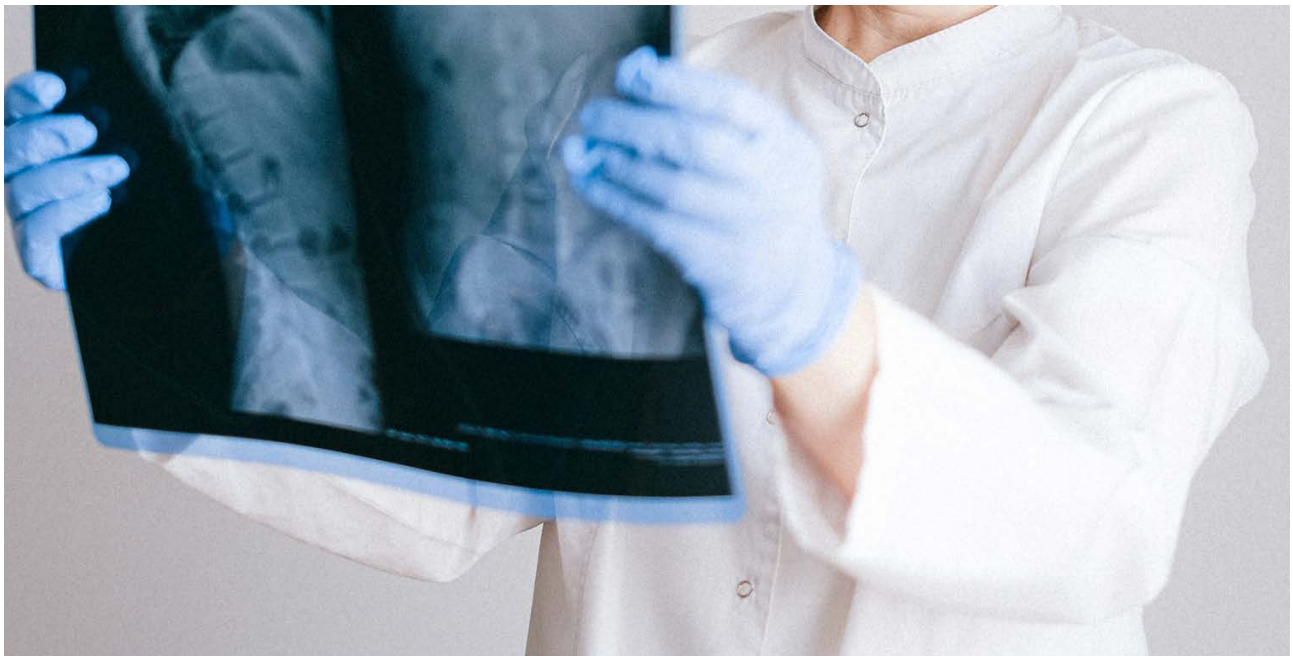


Table 2: Percentage of rare disease patients' access to treatment in Malaysia.[9]

Disease	% with treatment in Malaysia
Systemic lupus erythematosus (SLE)	NA
Spinal muscular atrophy (SMA)	77.8%
Osteogenesis imperfecta (OI)	97.8%
Maple Syrup Urine Disease (MSUD)	100%
Citrullinemia type II	9.1%
Duchenne Muscular Dystrophy (DMD)	88.5%
Marfan syndrome	50.6%
Prader-Willi syndrome	16.7%
Noonan syndrome	37.5%
Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes (MELAS)	58.9%
Leigh syndrome cause by mutations of nuclear genes	68.8%
Myoclonic epilepsy associated with ragged-red fibres (MERRF)	58.3%
Hunter syndrome or Mucopolysaccharidosis type II (MPS II)	23.1%
Pompe disease or Glycogen storage disease type II (GSD II)	58.8%
Multiple Sclerosis	87.5%
Cystic Fibrosis	44.4%
Haemophilia A	100%
Primary Pulmonary Arterial Hypertension	100%
Urea Cycle Disorders	62%
Fabry Disease	0%

3.1 Lack of treatment options

Only 10% of the known rare diseases have specific therapeutic treatment options. The rest rely on symptomatic treatment.

> ***P15: "So basically we need to identify what are the newer available treatments and whether everybody is supportive of the treatments; and you need some special programme for targeted rare disease, then you will know if the treatments have a positive impact for the patients and families."***

There are many factors that contribute to the lack of treatment options. A significant barrier to treatment development includes insufficient knowledge and understanding about rare diseases. The rarity of these conditions translates into a deficiency in terms of comprehensive scientific understanding, diagnostic tools, and treatment pathways. This knowledge gap can discourage pharmaceutical companies from pursuing research in these areas, as the complexities of these diseases can make drug development more challenging and uncertain.[37]

Developing new drugs, especially those targeting rare diseases, requires significant investment in research, clinical trials, regulatory approvals, and manufacturing. The costs associated with these processes can be substantial, and without a sizeable patient base, it can be difficult to recoup these investments and generate profits.[27] A small population size also complicates drug registration as randomised clinical trials would typically require large sample sizes. For instance, detecting a doubling in an adverse event that occurs at a rate of 1/1000 would require a sample size of 50,000.

With a small market size and steep development costs, pharmaceutical companies lack the incentive to develop drugs for rare diseases. The muted interest in drug development has given rise to the term "orphan drug" for drugs that are used to treat rare diseases. While there is no universal definition for "orphan drug" and "rare disease", the latter has been used to describe neglected disease by doctors and diseases that affect only a small number of individuals. Thus, an "orphan drug" can be defined as one that is used to treat an orphan disease.[38] The Ministry of Health uses the term "orphan medicine" and defines it as a "medicinal product that is primarily intended to treat, prevent or diagnose a rare disease" and is categorised either for emergency or lifetime treatment.

The combination of these factors has created a cycle whereby the scarcity of treatment options for rare disease patients persists hitherto. The lack of investment from pharmaceutical companies limits the development of new therapies, leaving patients with few or no effective treatments. This emphasises the importance of government initiatives, research institutions, and patient advocacy groups in driving efforts to fill the gaps in treatment options to help rare disease patients gain better access to treatment.

Patient Sharing #1

Beyond Skin-Deep: Jia Yun's Journey with Epidermolysis Bullosa Simplex

At 2.2kg—well under a newborn's normal weight at birth—Jia Yun was born in Teluk Intan in 2019 with skin that flaked, peeled, and bled at the slightest touch. Unbeknownst to the attending nurses and medical staff, this newborn was grappling with Epidermolysis Bullosa Simplex (EBS), a condition that leads to fragile skin and life-threatening blisters within the skin's epidermal layer. This rare condition has no cure. While EBS was undiagnosed at that point of time, that moment marked the beginning of a physically, financially, and emotionally taxing journey for both the newborn and his family.

When Jia Yun was first diagnosed, which in fact required a transfer to a hospital in Ipoh to get a proper diagnosis, his mother was clueless as to how to look after her baby. With no specialist nurse adept in Epidermolysis Bullosa treatment, she was "really hopeless" as the dermatologist available could only inform her of basic precautions, or the need to apply Vaseline periodically. Worse yet, Jia Yun was isolated from the other newborns at the NICU (Neonatal Intensive Care Unit). Witnessing this inflicted further misery and helplessness upon his mother.

The uncertainty and challenges they would eventually face were manifold. While newborn babies with EB can be effectively breastfed with many benefits, it proved to be a painful process as the skin around Jia Yun's lips would peel off during feeding. Handling and changing wound dressing would take almost 4 hours on end. Jia Yun would still cry from pain due to the numerous blisters and wounds, even after a small dose of morphine was induced for pain relief.

As Jia Yun grew older, the family's journey was fraught with a new set of tribulations. Any slight movement from Jia Yun—whether rubbing or scratching—would cause irritation and potentially, blisters. As such, his motor skills were delayed; only at the age of three did he start to walk firmly. Since his birth, Jia Yun's mother had no choice but to quit her job to care for him.

> ***"I was with him 24/7. I just stayed in the house. Even cooking and everything, my mother always sends [food] to me. I always make sure everything is clean [sic] with Dettol."***

Regular follow-ups with a paediatric physician also revealed that Jia Yun was potentially experiencing a delay in his cognitive development, which is not typical of EBS cases. Hence, his parents had to fork out a significant amount of money on occupational and speech therapy sessions, neither of which were covered by insurance or subsidies. Wound management also necessitated a lot of materials; from dressings, ointments, moisturising creams, alcohol swabs, normal saline, and other treatments, Jia Yun's parents had to financially sustain these expenses—all out of pocket.

> ***"We change the dressing three times per week. So, we buy lots of new dressings which is costly because one box has only 10 pieces. The supplier's cost is RM180. So, when the whole hand needs to be covered, it will need half of the box. I sometimes spend between 2k to 3k for dressing. One time, I bought 10 boxes in one month."***

DEBRA Malaysia, a support group, graciously arranged for Jia Yun's parents to purchase the dressings directly at supplier cost. Educational and moral support were also provided, all of which saw vast improvements in Jia Yun's health conditions.

The final, ongoing challenge faced by Jia Yun and his parents emerges on a societal level. Jia Yun was rejected by most kindergartens due to his condition. Out of 10 in the area, his mother approached 7, all of which rejected him except one. Today, Jia Yun attends kindergarten and is showing positive progress, all because he was given a chance.

Jia Yun's mother, as a caregiver to an EBS patient, aspires to create more awareness in the realm of education. From getting an accurate diagnosis, caring for his condition, affording said care, and ensuring that he gets a well-deserved childhood and education, Jia Yun and his mother's ongoing journey has been tough, to say the least. Yet, this story is just one of the many arduous paths that rare disease patients grapple with daily in Malaysia.

Note: The cost of dressings varies depending on the severity of the wound and the amount of dressing needed to manage it.

3.2 Costly treatment and diagnostic tools

Rare disease patients, saddled with the burden of their rare medical condition, also bear the high financial costs associated with accessing appropriate treatments and diagnostic tools. This poses significant challenges to the patients and their families in terms of disease management.

3.2.1 Cost of genetic screening

Genetic testing is an important tool for the diagnosis and prevention of lifetime impairment for many rare disease patients. Unfortunately, due to the limited local testing facilities, the specimen needs to be tested in an overseas laboratory. As genetic diseases can be hereditary, the test should also be expanded to the immediate family of the patient. It was estimated that the cost of genetic screening for diagnosing ANCA-negative Vasculitis is RM540 for the patient and RM1,800 for the parents. Testing for Marfan disease in a UK laboratory, a prevalent rare disease in Malaysia, can cost the patient RM1,700.

3.2.2 Cost of treatment

It was reported that orphan drugs cost roughly five times more than non-orphan drugs.[39] It was estimated that a paediatric patient on an Enzyme Replacement Therapy (ERT) requires between RM500,000 and RM1 million a year to sustain the cost of medication alone.[21] Furthermore, ERT is an ongoing treatment whereby its cost increases with age as the drug dose is administered according to the patient's weight. In addition, ERT cannot be stopped as the complications can recur. For example, idursulfase (ERT) for the treatment of MPS II costs approximately RM6,800 per vial. A patient who weighs 25kg would need two vials per week, at an estimated cost of more than RM700,000 every year. Other orphan drugs would cost up to RM 1.6 million per patient requiring ERT each year.[9]

Based on findings from the focus group discussions, it was reported that a patient suffering from ANCA-negative Vasculitis and receiving Immunotherapy Treatment (IVIG) needed to pay RM21,000, while a patient suffering from Achondroplasia who had two spine surgeries and three surgeries to correct bow-leggedness would incur costs of more than RM120,000. The cost of the metal needed in the surgery would be around RM2,000. In addition, a patient having a Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome may need a corrective vaginal surgery which can cost up to RM4,000. Prader-Willi syndrome (PWS) patients would need a continuous positive airway pressure (CPAP) machine or Bi-level positive airway pressure (BiPAP) machine to support their respiratory system, which costs RM20,000 and RM30,000 respectively.

Estimated Cost from Patient Sharing #1

The approximate annual cost of preventive treatment for a child suffering from Epidermolysis Bullosa Simplex (EBS) is as below.

- Cost of wound dressing: RM8,000
- Topical and moisturising creams: RM800
- Food supplement and vitamins: RM600
- Needles, alcohol swab, plasters, and normal saline: RM800

The total cost of daily consumables for health maintenance for 3 months: RM10,200

3.3 Ability to pay

The major challenge rare diseases patients face in utilising healthcare is their ability to pay for it. This refers to the capacity to generate economic resources—through income, savings, or loans—to pay for healthcare services without significant financial burden and impoverishment. On the other hand, affordability reflects the economic capacity for people to spend resources.

3.3.1 Government funding and limited financial support

Only drugs listed in the Ministry of Health Medicines Formulary List a.k.a. Blue Book will be readily funded by the government in the Ministry of Health facilities. It is very difficult for orphan drugs to pass the evaluation due to the extreme high costs and other related issues. For example, it was reported that the government only subsidises a few Enzyme Replacement Therapies, such as alglucosidase and idursulfase, as listed in the Ministry of Health Medicine Formulary.[9]

Hence, funding through the government most often comes from a case-by-case special approval channel, Ubat Kelulusan Khas (UKK) or New Programme Budget. The New Programme Budget is usually for specific diseases and centres, hence it needs to be continuously renewed to help patients gain access to optimal treatment. However, government funding for rare disease treatments is limited, and only a small number of patients who fulfil strict criteria have access to expensive therapy, primarily through hospitals run by the Ministry of Health.

In 2018, RM10 million was allocated for ERT treatment for rare disease patients. In 2019, Kuala Lumpur Hospital (HKL) was given RM16 million to treat rare diseases for six existing patients, whose treatment began in 2018, and eight new patients for 2019. The Ministry of Health allocated RM10 million in 2021 for follow-up treatments of existing rare disease patients and to commence treatments for patients on waiting lists. In 2020, Ministry of Health allocated RM16.5 million to treat patients with lysosomal storage disease, Prader Willi syndrome, cystinosis, and other rare diseases.[25]

In addition, patients with rare diseases also encounter barriers in accessing treatment and care in healthcare facilities managed by different ministries given the limited funding by the government.

> ***P4: "I've been treated in a teaching hospital. It is under KPT (Kementerian Pengajian Tinggi), it's not under KKM. So, I couldn't get the funds for IVIG. [Redacted] I cannot because ANCA vasculitis is [not] listed as a rare disease. Thus, I am not eligible."***

3.3.2 Out-of-pocket (OOP)

The financial burden incurred due to out-of-pocket treatment costs might render rare disease patients and their families economically vulnerable. Patients often find themselves spending potentially catastrophic amounts on treatment procedures (such as spine surgery or corrective surgery), diagnostic tests, materials, and supplements. These burdens can limit rare disease patients' access to healthcare services and affect their quality of life.

> ***P9: "Generally for PWS when they are diagnosed, because of the low muscle tone development, they will have a lot of respiratory problems. So, they have to be in the hospital all the time, for the first year. And before discharge, they need to ensure that they have a CPAP machine or BiPAP machine. The cost for [a] BiPAP machine is about RM22,000 and CPAP [is] probably lower than that. But during my time [the] CPAP cost was almost RM20,000, and [a] BiPAP was almost RM30,000 in the year 2000s. Okay, if you don't get that machine, you have to stay in the hospital."***
>
> ***P1: "From personal experience, my son had two spine surgery [sic] and three for bowlegged [sic]. I think until today, it cost me about more than RM120k."***

There is a wide range of OOP expenses by rare disease patients in Malaysia, ranging from RM1,600 annually for an adult patient with Systemic Lupus Erythematosus (SLE), to RM10,796 annually for an adult patient with ANCA-negative Vasculitis, and RM17,720 annually for a child patient with Epidermolysis Bullosa. An analysis of their OOP outpatient expenses shows that private clinic prescription drugs incur the largest cost (34.1%) followed by private clinic consultation fees (31.8%), over-the-counter medicine (22.7%) and supplements (11.4%). In addition to the OOP expenses, some patients require a full-time caregiver, professionally engaged and costing around RM3,500-RM5,000 monthly. Often, these caregivers are parents/relatives who have had to quit their job to attend to the patient's needs. This productivity cost increases the overall economic loss. Abu Bakar et al. (2020) reported the indirect costs (productivity) for SLE patients were RM2,875.17 over a period of seven days.[40]

In Europe, a 2017 survey by the European Organisation for Rare Diseases revealed that 15% of rare disease patients could not access treatment due to financial constraints.[41] Meanwhile, in China, a study in 2016 found that over 90% of rare disease patients faced financial challenges meeting their living expenses, and, on average, the medical costs for individuals with rare diseases were three times higher than their income.[42] Additionally, in Africa and the Middle East, rare disease patients face challenges due to the unavailability of orphan drugs and the lack of reimbursement systems for these drugs.[43]

Despite the universal healthcare coverage in Malaysia, patients and their caregivers are still financially burdened because many of the expensive treatments, diagnostic tests, materials, and supplements for rare diseases are not covered by the Malaysian public healthcare system. As rare diseases frequently require lifelong treatment and dependency on assistive devices and supplements, it represents a major share of the economic burden for rare disease patients.

> ***P15: "I think [the subject of] rare diseases is more complicated because there are so many different rare diseases. So, whatever is announced for rare diseases in the budget speech, that is only a portion of the money that the government spent on the rare diseases, especially for a few types of rare diseases only. Those rare diseases have some established treatment programmes but of course the money is not enough because there are more and more rare diseases that need specific treatments and they have yet to have established programmes. Whatever is announced in the budget speech is mainly for Lysosomal Storage Diseases (LSD) because those have long-established programmes. The newer diseases like Systemic Mastocytosis (SM), Cardiac Amyloidosis and [a] few others, they don't have an established programme."***

The management of diagnosis and treatment of rare diseases is also challenging. Providing care for rare disease patients is time intensive, forcing many caregivers to reduce or stop their professional activities, putting households under financial strain.

> ***P1: "Since my son was born and diagnosed with [a] rare disease condition, I changed jobs. I looked for a job that pays more and also has flexible hours so that I can attend to this unknown disease."***

A study conducted in Malaysia reported that 43.2% of SLE patients experienced work disability due to SLE-related factors, such as renal manifestations and organ damage.[41] Meanwhile in Australia, a study revealed that 45% of parents to children with rare diseases struggle to manage the associated costs, which leaves 29% of them having to increase their working hours or take on second jobs.[44] Also, in Canada, caregivers have reported financial vulnerability, with 63% accumulating debt to cover caregiving expenses.[45] In Europe, a survey conducted in 42 countries confirms the significant impact of rare diseases on the well-being of patients and their caregivers.[46] According to the findings, 70% of patients and their caregivers had to reduce or halt their professional activities due to the disease, with 21% absent from work for over 90 days annually. Hence, funding and financial support is one of the most urgent means to enhance the lives of rare disease patients.

4.0 HEALTHCARE CONSEQUENCES

4.1 Despair due to limited treatment options

Patients' engagement in healthcare is strongly determined by their capacity to participate in the care and management of their condition. However, due to the limited treatment options, patients often find themselves unable to self-manage their condition or comply with treatment management.

> ***P7: "Previously, when the doctor says, Spinal Muscular Atrophy (SMA) has no cure, there was nothing to do [sic] except physiotherapy. So I've been just doing physiotherapy since then. However, for some rare disease patients, they just give up going to the doctors, because maybe the doctor says this is hard to get a cure or to have a treatment unless it's just physiotherapy that you can do at home. So the patients just give up."***

A study conducted in the United States identified that the lack of available treatment was one of the main reasons why 1 out of every 3 caregivers would have difficulties in accessing treatment for symptom management or rare disease treatment.[47] This will eventually cause patients to give up on treatment. As a result, adverse psychological and emotional impacts on patients and caregivers were often reported. Based on findings from the focus group discussions, rare disease patients and their families are at higher risk of experiencing poor quality of life, increased mental health issues, more social isolation, poor work-life balance, and discrimination in employment.

> ***P8: "This is very crucial if you are employed with the government offices because you have to get yourself confirmed from the government hospitals that you are fit to work. Most of the time, they will not approve that we can work. Another example is [a] rare disease person (with albinism), she was a training [sic] teacher who is [sic] applying for a confirmed position as a teacher in Sarawak. She got rejected, so that's the kind of discrimination in employment."***

Patient support groups could help patients facing employment discrimination by advocating for a more favourable work policy environment for rare disease individuals, encouraging inclusive workplace policies, and educating employers about their capabilities and needs. By engaging in open conversations, partnering with support networks, and sharing personal stories, individuals with rare diseases can help reshape perceptions, promote diversity, and ultimately create more inclusive work environments that accommodate their unique needs.

4.2 Poor services transition

It is important that there is an appropriate fit between the services provided and the patient's needs. Based on findings from the focus group discussions and roundtable, it was highlighted that there is a poor healthcare transition between paediatric and adult clinics are due to the lack of adult rare disease subspecialists.

> ***P16: "Now we have a lot of survivors of rare conditions, they are treated, and now they are 21, 30 and 40 years old. We [as healthcare providers] tried to refer them to adult services, but very often they just come back and say "I don't want to go there, I want to stay here." So you'll find big adults in a paediatric clinic. Therefore, now we have to change our perception [of] this cohort of patients that have been surviving and are now moving into the realm of adulthood. And they are now facing problems that their predecessor[s] had never faced before. Like, what do you do with jobs? What career can they have? Can they get married? Do they want kids? And they want choices. They want all sorts of things as a citizen of [the] country and to be productive. So, this big group of patients who are like, if you have neglected them, so they will be like [the] disabled and we will have an issue. But if you look after them properly, they are capable, they can actually contribute as productive citizens of [the] country. But unfortunately, they can't transition properly. For example, the healthcare [system] is not equipped to deal with adults with rare conditions. We still see all these adult patients [in a paediatric clinic] who have rare conditions because they have nowhere to go and nobody wants them to be referred."***
>
> ***P15: "Now they are adults, so they need to have some shelter workshop [sic] certain job placements. That's the question we're frequently hearing from parents. Now they're sitting at home because the Special School is only until 19 years old. Some of them can do some work but there is no way that they can find a centre that can accommodate them."***

Hence, the healthcare landscape needs to evolve in response to the changing needs of this growing cohort of rare disease patients. The shift from paediatric to adult care presents a unique set of challenges, where patients often encounter a lack of appropriate services and support systems tailored to their adult needs. This transition encompasses not only medical care but also broader life aspects such as employment, career choices, marriage, and independence.

To address these issues and create a more inclusive society, it is important for healthcare systems to bridge the gap between paediatric and adult care, offering specialised services for adults with rare conditions. This includes establishing shelter workshops, job placements, and centres that cater to their specific requirements, ensuring that these individuals can lead fulfilling lives as productive citizens. Collaborative efforts between healthcare providers, social services, and advocacy groups are essential to create a comprehensive support network that empowers rare disease survivors to overcome challenges and contribute positively to their communities.

Policy Recommendations

Having identified the key barriers throughout the patient pathway, this study calls for four critical pillars of reform to improve healthcare access for rare disease patients. While there are many challenges which are likely impossible to address holistically in this paper alone, these policy recommendations focus on tangible changes that can be made to address recurring and immediate problems faced by rare disease patients. Thematic content analysis was employed to generate codes and themes from data obtained during focus group discussions with patients and a roundtable session with stakeholders. The themes obtained were then mapped onto Levesque's framework to highlight the key barriers and challenges to accessing optimal treatment and to finalise the selection of policy recommendations. However, care must be taken to acknowledge the precedence of patients' perspectives in this paper. As our findings have been extrapolated from a series of engagements with patients, these recommendations are from a patient-centric viewpoint and suggest policy measures that can alleviate their most pressing concerns.

Based on the key barriers highlighted throughout the patient pathway, this study calls for four critical pillars of reform to improve healthcare access for rare disease patients and stratify them based on their complexities (see Table 3). Strategies to remedy root causes for issues that cut across the journey path are considered strategic pillars. Among the elicited issues, limited rare disease specific healthcare resources was found to have a significant impact on patient's health care seeking, reaching, and utilisation activities.

Having limited diagnostic equipment and healthcare professionals results in delayed diagnosis for the patient. They also necessitate service centralisation, that in return increases patients' out-of-pocket cost, reduces their productivity and increases treatment attrition. In this section we propose an action plan of policy recommendations that could be implemented in the short-term, mid-term, and long-term to directly address barriers along the rare disease patient pathway.

Table 3: A Multidimensional Policy Framework on health equity for individuals with rare diseases and their caregivers.

Strategic Pillars	Aspects of Policy Development	Timeframe
 **Pillar 1: Enhancing healthcare resources capacity**	Building the capability of healthcare professionals through collaboration, strengthening the role of universities and prioritising government position warrants	Short-term *(1-2 years)*
	Resource sharing for diagnostic tests between interministerial facilities	
	Improving seamless collaboration and integration between healthcare facilities and patient groups	
 **Pillar 2: Comprehensive and sustainable rare disease financing**	Cultivating the orphan drugs market through regulatory incentives	Mid-term *(3-4 years)*
	Ensuring affordability of treatment through risk sharing mechanisms	
	Leveraging pooled procurement to boost purchasing power	
	Accelerating the Health Technological Assessment (HTA) process through joint assessment	
	Strengthening the role of the trust fund to address unmet needs of rare disease treatment affordability	
	Developing the pharmaceutical industry to promote generic competition for drugs treating rare diseases	
 **Pillar 3: Strengthening governance for holistic management of rare diseases**	Institutionalising a legal framework for rare diseases to protect patients from heightened vulnerability	
	Interministerial coordination and collaboration as key to improving integration and continuity of care	
 **Pillar 4: Enhancing social protection systems to promote resilience of individuals with rare diseases and their caregivers**	Protecting rare disease patients and householders with improved insurance coverage	Long-term *(more than 5 years)*
	Improved coverage and assistance for support services and assistive devices	
	Promoting inclusive employment opportunities for people living with rare diseases and their caregivers	

Patient Sharing #2

Lost in Transition: SLE Misdiagnosis and Healthcare Inaccessibility

From 2000 to 2006, Sophia was working in Singapore when she started to experience swelling in her leg and frequent ankle pain. While these were the first symptoms of Systemic Lupus Erythematosus (SLE), a rare inflammatory autoimmune disorder, her initial diagnosis at age 30 indicated that she was suffering from mixed connective tissue disease (MCTD). Educated on MCTD symptoms and its management at a hospital in Singapore, Sophia only heard of SLE or lupus mentioned in passing.

It was only upon her return to Malaysia in 2006 that she knew of her SLE diagnosis enclosed within her referral letter from Singapore. With SLE, the immune system mistakes healthy parts of the body as foreign and attacks them, causing inflammation and tissue damage. Failure to adhere to treatment leaves SLE patients at high risk of kidney failure. Despite the gravity of her condition, she was left in the dark about her final diagnosis, only discovering the truth through her reference letter.

> ***"I asked for a reference letter so that I can continue my medication. At that time, I was still thinking that I had MCTD. However, in the letter, it mentioned lupus. So, I didn't know actually! I remember the nurse told me that it's not easy to be diagnosed with lupus, it takes time because you have to do tests to confirm it. So that's why they diagnosed me with MCTD first. I only knew that I had lupus when I had that referral letter."***

In the early stages of her journey with SLE, Sophia had no access to support groups, nor any education on what SLE was. No one around her could offer an explanation, which she recalls as incredibly frustrating. When receiving regular checkups, Sophia was unable to access her test results and physician's report, which meant she could not ascertain if her condition was improving or to what extent her lifestyle changes contributed to this improvement. While the doctors would repeatedly say that everything is *"okay"*, the inability to have a copy of her results left Sophia oblivious to the progress of her condition.

Sophia also highlighted how Malaysian healthcare differed vastly from that of Singapore in terms of accessibility to medical records across hospitals. While Singapore boasts an integrated system, where access to her medical record and previous meeting transcripts were available across different hospitals or polyclinics, this was not the case in Malaysia.

> ***"In Malaysia, every time I go and see the doctor, the doctor will ask me the same question like: What do you have? Where is the pain? When were you diagnosed? You know all these basic things will be [sic] asked to me when I first came to the MOH hospital, which frustrates me."*** *(The patient has since clarified that as of now, this no longer happens).*

The transition from Singapore to Malaysia also incurred a financial barrier. Despite being a Malaysian citizen, Sophia faced difficulties accessing free treatment as her referral letter was from a Singaporean hospital. When receiving treatment at the MOH hospital, she had to bear the cost of all of her medication and blood tests out of pocket, on top of being ineligible for an OKU card as she was able-bodied.

As a low-income individual, saddled with the burden of frequent hospital visits and blood tests, Sophia eventually encountered legal trouble due to her inability to pay for her treatment expenses. Legal letters wound up at her doorstep due to the accrued expenses that she failed to pay in full. Only after repeated appeals for payment exemption did she obtain approval for free treatment.

Today, Sophia pleads for more support and education for SLE patients from the Malaysian healthcare system. Specifically, she yearns for the establishment of customised consultation sessions where healthcare professionals can answer patients' questions regarding their condition, regardless of its rarity.

We can no longer leave the responsibility of rare disease management to be borne solely by the patient, but rather it is the shared duty of medical professionals and society as a whole. Sophia's story is a wake-up call for the Malaysian healthcare system to work towards better doctor to patient communication and heightened integration across healthcare institutions.

PILLAR 1: ENHANCING HEALTHCARE RESOURCE CAPACITY

There is an urgent need to enhance the local diagnostic, treatment, management, and education capability of rare diseases in Malaysia. Delayed diagnosis can lead to disease mismanagement, with dire consequences on patients' morbidity and mortality.

1.1 Building the capability of healthcare professionals

To sustainably enhance service delivery, more emphasis should be given to foster the skills and knowledge of healthcare professionals. Whilst rare disease patients are primarily treated by paediatric clinical genetic specialists, many of their complications require multiple clinical discipline specialties. For example, Marfan disease patients often require cardiologists to monitor the weakening of the aorta which is common in the disease.

Clinical audits can be conducted to identify the gaps in the healthcare profession. Likewise, systematic training curricula for the pre- and in-service healthcare professionals can be developed through close collaboration between medical facilities and training universities, and between private and public sectors. The training ground can also be further expanded by setting up a regional or international network that promotes the exchange of knowledge and experience, ideally regarding the specific disease groups.

Many of the issues in rare diseases can be addressed effectively and efficiently through an interdisciplinary (medical and non-medical) approach. Whilst the roles of clinicians are well understood and charting good progress, other professionals such as pharmacists, epidemiologists, lawyers, and policy experts also have a crucial role to play in improving the process of orphan product approval and management. Early interactions and experiential learning with the other professionals on rare diseases will improve understanding of, and help find solutions to, existing rare disease issues.

Universities with teaching hospitals can be an effective platform to support the training of multidisciplinary professionals. The pool of expertise, patient base, and multidisciplinary faculties will allow a richer learning experience while simultaneously optimising patient treatment.

Given the importance of these universities in the capability building process, they need to be sufficiently funded in order to maintain and strengthen the current ecosystem. An inter-ministerial mechanism should be established to allow the cross-funding of patients between the Ministry of Health and Ministry of Higher Education facilities. The Ministry of Higher Education can also collaborate with the Ministry of Health, Ministry of Women, Family and Community Development, and the Ministry of Science, Technology and Innovation to establish research and training funds to provide innovative solutions to rare disease issues and to train more professionals.

One major issue occurring in many newly established government healthcare institutions is the lack of positions for sub-specialists. For example, some of the fully trained genetic counsellors are not able to practise professionally due to the lack of such positions in genetic clinics. Hence, the Ministry of Health should continuously engage with the Public Service Department to allow a proper allocation of positions for allied healthcare professionals that support the function of genetic clinics. This concern also extends beyond the scope of genetics alone, as in some cases, fully qualified specialists are unable to practise due to the absence of such sub-specialities in the public healthcare system. Assigning a higher priority for rare disease related specialties in their warrant allocation will ensure equitable access for these patients.

1.2 Resource sharing for diagnostic tests

Advancements in molecular genetics over the last few decades allow for better characterisation of the causes behind many rare diseases and provide unprecedented opportunities for diagnosing individuals and determining phenotypes. However, genome sequencing and lab capacity in general is limited and still unaffordable in Malaysia.

Malaysia can learn from the recent experience it had during the COVID-19 pandemic, where resources were pooled across different ministries for collecting, storing, and analysing specimens for diagnosis and serotyping. The country could establish a national network to build and share genetic testing and diagnostic infrastructure, therefore leveraging each institution's strengths. The network could be extended to regional platforms through selected bilateral and multilateral collaborations, and organised based on their respective economies.

Take for example, the Analytical Biochemistry Research Centre of Universiti Sains Malaysia has the technical and infrastructure means to screen for inborn error metabolism using Tandem Mass Spectrometry. As the Ministry of Health has previously engaged this service, the collaboration can be

reactivated to support the newly established genetic clinic service for the northeast region located in the Hospital Pulau Pinang.

1.3 Improving seamless collaboration and integration between healthcare facilities and patient groups

The psychological and knowledge-based support given to rare disease patients and their caregivers thus far have mainly stemmed from NGOs and patient support groups. Clinical practitioners, contrary to expectations, largely lack rare disease-related expertise and awareness. As a result, most rare disease patients grapple with delayed diagnosis or experience misdiagnosis.

While NGOs and patient groups—namely, Persatuan SLE Malaysia (as mentioned by our interviewee), Rare Diseases Alliance Foundation Malaysia (RDAFM), and Malaysian Rare Disorders Society (MRDS)—do indeed helm the social and psychological support of rare disease patients in Malaysia, this is gravely insufficient without the clinical expertise of practitioners and specialists. To bridge this disconnect, we suggest adopting the European Reference Network (ERN)[48] as a model to establish a National Rare Disease Network to pool and facilitate the mobility of expertise. These networks should also act as a patient-navigation platform to link healthcare facilities with other stakeholders such as patient groups, volunteer organisations, research institutions and diagnostic laboratories across the country and internationally to ensure the exchange of, and equal access to scarce knowledge.

> ***"These networks should also act as a patient-navigation platform to link healthcare facilities with other stakeholders."***

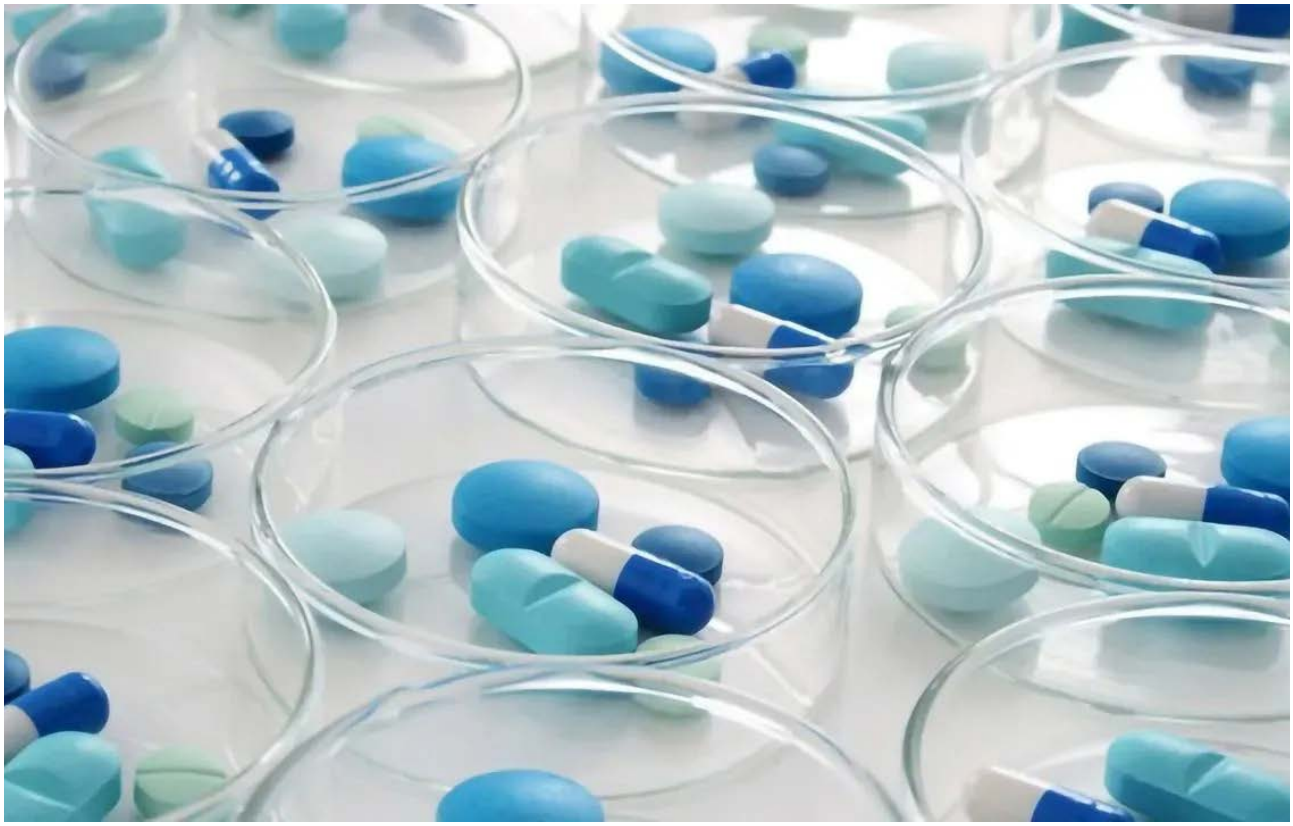
Upon encountering cases of rare diseases, this network can enable healthcare professionals to discuss a patient's diagnosis with relevant stakeholders via an online platform. By equipping healthcare professionals with knowledge previously inaccessible to them, the quality of integrated care for rare disease patients can be enhanced through appropriate and timely diagnosis.

PILLAR 2: COMPREHENSIVE AND SUSTAINABLE RARE DISEASE FINANCING

A policy and an operational framework within the healthcare system that drives innovation while ensuring affordability for patients is ideal. However, when considering policy proposals addressing rare disease products, there are trade-offs between measures taken to improve affordability and innovators' incentives for new drug development. As such, in considering legislations and policies that favour orphan drugs, it is equally important to also take into account the policies that aim to ensure the affordability of rare disease treatment.

2.1 Cultivating the orphan drugs market through regulatory incentives

The inherent risk associated with orphan drug development and the very small patient population which rare disease treatment could derive revenue from are key barriers limiting the availability of specific drugs for many of the rare diseases.



> ***"In order to create a viable business model for rare disease treatment, regulation is needed to correct the structural imbalance in the risk and reward for rare disease treatment."***

For instance, the United States government legislated the Orphan Drug Act (1983), with the aim to correct structural imbalances in the risk and reward for rare disease treatment. Among other incentives (see Table 4), this act provides tax credits to offset some of manufacturers' research and development costs, and eligible products receive an extended seven-year market exclusivity. Since its passage, there has been an increase in the number of products approved to treat rare diseases, with over 800 orphan drug indications approved between 1983 and 2019.[49]

Many approved orphan drugs have contributed to improved treatment in the oncology space, with 42% of the 491 orphan drugs approved over the past ten years targeting rare cancers. In Europe, a comparable regulation was implemented, which resulted in the introduction of 65 orphan drugs between January 2010 and July 2011. In Asia, South Korea listed 58 orphan drugs over seven years (2010 to 2013) through the National Health Insurance Corporation negotiation process.

It is also important to note that this progress has also been spurred by advances in cell and gene therapies, among other scientific discoveries, that have revolutionised care for many rare conditions. The patient community has contributed to this success and has demonstrated its ability to spur investment, inform research and influence policymakers at all levels.

Following the publication of its first Malaysian Orphan Medicines Guidelines in 2021, Malaysia has introduced the accelerated pathway for registering designated orphan drugs.[50,51] The pathway allows the registration of orphan drugs to be prioritised with a more flexible clinical evidence requirement. This will reduce the costly development process for the manufacturer and serve as an incentive for more orphan drugs to be developed and marketed in the Malaysian market. At present, there are only two drugs (Enspryng™ 120mg and Koselugo® 25mg) that have received approval to be designated as orphan medicines. These medications are used to treat Neuromyelitis Optica Spectrum Disorder and neurofibromatosis respectively.

Malaysia could take a step further in the regulatory process through the legislation of a formal rare disease act that could offer more incentives for drug developers, and strengthen interministerial governance. These incentives could then be used to leverage its price negotiation in government facilities. Without the protection of such legislations, rare disease patients in Malaysia depend heavily on federal budget and ad-hoc allocations, as well as crowdfunding through patient-advocacy groups, in order to access orphan drugs. With less than five orphan drugs approved in Malaysia, and a large number of rare disease patients not having treatments made available to them, the unmet needs of patients and families across many rare disease areas in Malaysia remain extremely high.

Table 4: Economic incentives created from the Orphan Drug Act (1983) in the United States.[52]

Policies or Programmes	Description
Longer market exclusivity	7 years of market exclusivity for approved orphan indications
Tax credits for expenditures incurred in conducting clinical trials	25% federal tax credit for expenditures incurred in conducting clinical research within the US
User fee waiver	Waiver of Prescription Drug User Fee Act fees
Research grants	Ability to compete for research grants from the Office of Orphan Products Development (OOPD) to support clinical studies for orphan drugs

2.2 Ensuring affordability of innovative rare disease treatments through risk sharing mechanisms

While such legislation has shown to generate rapid growth in approved rare disease treatments in recent years, it also comes with a very high cost.

> ***"To ensure that rare disease treatment is sustainable and equitable, policies cannot just be about creating a viable business model, but rather also targeted at absorbing the costs of the growing wave of high-priced orphan drugs."***

There are a range of potential policy reforms that could be implemented in tandem with regulatory incentives for orphan drugs, in order to ensure affordability of such treatments:

Table 5: Potential drug pricing policy instrument to address the high prices of rare disease treatment products. [51]

Policies or Programmes	Description
Outcome-based contracts (OBC)	OBC makes some or all of the payment of a treatment contingent on the degree of patient benefit. Such a model could take several forms, with sliding scale bonuses or refunds depending on outcomes. These contracts require manufacturers and payers to agree on a set of measurable outcomes and to track those outcomes in order to adjudicate the contract. Though such efforts have shown to be administratively burdensome and expensive to negotiate and implement, it has the benefit of addressing the interlocked concerns about the pricing of orphan drugs with those regarding the uncertainty of the evidence on their effectiveness.
Volume-based contracts (VBC)	VBC is an approach that could support rare product commercialisation by guaranteeing coverage, promoting patient and provider education, assuring equitable access and utilisation and simplifying contracting. While this model has historically been used by the government to purchase a large volume of drugs such as vaccines, this model can also be adapted for orphan drugs. To achieve the necessary product volumes for rare conditions at an affordable price requires a single-purchaser model. Under this model, the federal government or a consortium of private payers could directly negotiate to purchase enough orphan product volume to cover all eligible patients with a given rare condition. The contract would enable the government or private consortium to set a price unique to orphan drug indication. Manufacturers would benefit from a single contract, improved patient access and predictable utilisation. These contracts could also be structured to ensure that product prices fall as utilisation expands.
Indication-based pricing (IBP)	IBP enables payers to negotiate higher prices for rare indications and lower prices for broader indications or those for which the product demonstrates less clinical value. Independent value assessment entities could help establish a value-based price for a given rare condition and indication, based on the clinical benefit and strength of the evidence by disease area. While indication-based pricing would help expand access, one potential risk is that this pricing flexibility would allow manufacturers to increase prices for high-value indications, which could increase cost-sharing for rare disease patients.
Value-based price regulation (VPR)	Value-based price regulation is a mechanism that sets the price based on society value of health (cost-effectiveness threshold). The price then could shift over time as additional evidence is generated. As such, value-based price setting creates incentives for investment in evidence development to demonstrate the clinical benefits to patients, while scaling those rewards down for drugs that do little to improve patient outcomes.

In Malaysia, these risk-sharing mechanisms are referred to as the Patient Access Scheme (PASc). PASc, otherwise known as managed entry agreement (MEA), is a type of conditional agreement between the buyer and the manufacturer that aims to facilitate access to expensive drugs.[53] There are many forms of MEA such as price-volume capping, outcome-based agreements, rebates, discounts, paybacks, risk-sharing agreements or performance-based agreements. The aim of these agreements is mainly to control the budget expenditure and to address the various uncertainties regarding the effectiveness of treatment.[54] At present, no PASc exists for any of the orphan drugs in Malaysia.

Italy has been one of the leading countries incorporating MEA in the process of price negotiations for pharmaceutical products and has successfully achieved cost savings while increasing access to innovative medicines for its population. Based on Ashraf and Ong (2023),[55] there are a few policy recommendations that we can draw from Italy's MEA in formulating Malaysia's PASc:

1. Improving data infrastructure (i.e. data registry) to monitor real-world effectiveness of drugs—an integral part of drug pricing and reimbursement.

2. Establishing an independent regulatory body that is committed to continuously engaging with pharmaceutical manufacturers through continuous discussion to reach an agreement.

3. Formalising the terms of agreement in the price negotiation process between the regulator and manufacturer to reduce unintended risks.

4. Implementing various forms of risk-sharing and payment mechanisms in the agreement which ensures that the pricing framework remains flexible and accommodative.

2.3 Leveraging pooled procurement to boost purchasing power

Pooled procurement of medicines is a possible solution to reduce prices, increase availability, and achieve more efficient procurement processes. It can be defined as a collaborative initiative for buyers to consolidate their medicine purchases. The mechanism can be traced back to the late 1970s and can be used to improve affordability for the government to purchase orphan drugs.

> ***"In the longer term, the government should develop a platform to procure drugs on a regional basis in order to leverage broader purchasing power. "***

2.3.1 Interministerial

In Malaysia, drugs are procured by each ministry separately, and an internal inquiry found a marked difference in the prices paid for drugs by each ministry. A pilot project initiated in collaboration between the Ministry of Health, Ministry of Defence, and Ministry of Higher Education to procure 88 items through pooled procurement showed a total saving of 17.7% (RM179 million) to the government compared to individual ministry purchases.[56] In fact, there is a marked reduction between 2.9-27.5% of the median drug price with pooled procurement.

A similar approach may be taken to purchase drugs for rare disease patients as some are being seen in university hospitals instead of Ministry of Health's facilities. This will boost purchasing power and lower the cost to the government to procure and provide more care to rare disease patients.

2.3.2 Regional

Pooled procurement of medicines and strategic health supplies at regional levels has been successfully applied by the Gulf Cooperation Council (GCC), PAHO Revolving Fund (RF), Global Fund to Fight Aids, Tuberculosis and Malaria (Global Fund), the Global Drug Facility (GDF), and the President's Emergency Plan for AIDS Relief (PEPFAR). The most recent initiative, in which Malaysia also took part, was the pooled procurement initiative to obtain vaccines and personal protective equipment in the fight against COVID-19.

Regional procurement allows individual governments to address current challenges on a national scale such as irregular releases of government funds, budget shortfalls, limited in-country capabilities in procurement and management, and complex or lack of transparent procurement practices. The mechanism was found to increase competition among suppliers and consequently reduce prices between 70–82%. It also increases equity by providing uniform pricing to all member countries, irrespective of their market size or level of development. Transparency was also increased in the procurement process, improving the regularity of supply of essential commodities to countries and allowing countries to introduce newer, more expensive products.

Taking the example of PAHO, participating countries delegate to the agency the authority to conduct, on their behalf, most aspects of vaccine procurement—from issuing tenders to contracting with and paying suppliers. Prerequisite to the procurement is a common, revolving fund that allows PAHO to pay producers before countries reimburse the funds upon acceptance of goods in-country.

Regional procurement tools however require a collective agreement, with different economic status among members of Association of Southeast Asian Nations (ASEAN), on definitions of orphan drug or rare diseases and commitment from the respective governments for funds, infrastructure, and legal framework. This can be solved at regional level through either the APEC or ASEAN platform.

2.4 Accelerating the Health Technological Assessment (HTA) process through joint assessment

The process of Health Technological Assessments (HTAs) should be more directly integrated with price negotiations in order to leverage the clinical and economic assessment undertaken as part of the HTA. The government could consider options to press its negotiating position, beyond the current PASc guidelines. For example, the government could offer producers of specific treatments exclusivity for their treatments in the public health system beyond the patent term, in return for a reduction in the unit price of the treatment.

A more sophisticated use of HTA however requires improved resources, including technical expertise and evidence-generation ecosystems. However, the HTA agency and committee in Malaysia severely lack such capacity.[57] A comprehensive HTA also requires robust fundamental epidemiology, clinical, and economic evidence, which is unfortunately often missing in Malaysia. Consequently, there is a limited scope and number of HTA that can be conducted in Malaysia. Building internal capacity requires strong and long-term commitment and investment in both the capacity and the ecosystem. As an intermediate measure, harnessing regional evidence and expertise will enhance the country's HTA capabilities.

Developing internal capacity necessitates a steadfast and sustained commitment, coupled with substantial investments in both the capacity itself and the supporting ecosystem. As an interim measure, leveraging regional evidence and expertise presents an opportunity to enhance the nation's HTA capabilities.

An example of this model is the Beneluxa initiative in 2018 (see Table 6), whereby Belgium, the Netherlands, Luxembourg, Austria, and Ireland established a cooperative initiative on pricing and reimbursement, including the joint negotiations for drugs. The case of Beneluxa demonstrated that countries with similar levels of income demonstrate presumably similar healthcare spending appetites; this supports the feasibility of a regional cooperation and makes a clear case for Malaysia to advocate and participate in such a platform. ASEAN has shown some commitment to understanding the capacity and needs of ASEAN Member States to institutionalise HTA.[58]

Table 6: Joint Health Technology Assessment (HTA) procedures that were carried out and performed according to national legislation by the Beneluxa Initiative on Pharmaceutical Policy.[59]

Branded Name	Company	Therapeutic area by European Medicines Agency (EMA)	Year	Type of HTA collaboration
Lojuxta	Aegerion	Hyper-cholesterolemia	2015	Belgium re-used Dutch HTA work
Orkambi	Vertex	Cystic fibrosis	2016	First submission a. Joint HTA between Belgium and Netherlands b. Dutch Zorginstituut as external referee for RIZIV-INAMI c. Luxembourg used the final report.
			2017	Second submission a. Joint HTA between Belgium and Netherlands b. Dutch Zorginstituut as external referee for RIZIV-INAMI c. Final report sent to Luxembourg and Austria
Praluent	Sanofi	Dyslipidemias	2016	Dutch Zorginstituut acted as external referee for Belgium RIZIV-INAMI
Vyndaqel	Pfizer	Amyloidosis	2017	a. Dutch Zorginstituut as external referee for RIZIV-INAMI b. Luxembourg used the final report.

Ocaliva	Intercept	Amyloidosis	2018	Joint HTA between Belgium and Netherlands
Spinraza	Biogen	Spinal Muscular Atrophy	2018	Joint HTA between Belgium and Netherlands

Within Malaysia itself, while there is some form of institutionalisation of HTA, there is no formal legislative mandate and a lack of overall support for the uptake of HTA in health policy (see Table 7).[60] An effective national HTA implementation can work hand-in-hand with expanding regional political capital. Pursuing cooperation with regional networks such as the Asia Pacific Economic cooperation can leverage feasible mechanisms moving forward.[61]

Table 7: Legislations for HTA in Southeast Asia. [61]

Country	Legislative Mandate	Other legal provisions	HTA nodal agency
Malaysia	No	Yes	Malaysian Health Technology Assessment Section (MaHTAS)
Indonesia	No	Yes	Indonesian Health Technology and Assessment Committee (InaHTAC)
Lao PDR	No	No	Unit for Health Evidence and Policy (UHEP) Will be established to serve as focal agency for all HTA activities
Myanmar	No	No	–
Singapore	No	Yes	Agency for Care Effectiveness (ACE)
Thailand	No	Yes	Health Intervention and Technology Assessment Program (HITAP)
The Philippines	Yes	No	The Department of Health's Health Technology Assessment Unit (HTAU)
Vietnam	No	Yes	Health Strategy and Policy Institute (HSPI) A unit dedicated to HTA will be established soon

2.5 Strengthening the role of the trust fund to address unmet needs of rare disease treatment affordability

> ***"To further address the unmet needs by increasing the budget for rare disease treatment, a ring-fenced funding model can be considered."***

Some notable examples of such funds are the New Medicine Fund in Scotland as well as Singapore's Rare Disease Fund, both of which were established to provide financial support for rare disease treatment (see Table 8). Though not specific to rare diseases, the Bhutan Health Trust Fund is also a general health trust fund which adopts a similar model with different funding sources.

Table 8: Comparisons of health trust funds between Singapore, Scotland and Bhutan.

Country	Rare Disease Trust Fund (Singapore)	New Medicine Fund (Scotland)	Health Trust Fund (Bhutan)
Fund size	$70 million (18 million raised)[62]	£50 million (2021-2022)	$42.9 million[63]
Fund type	Endowment: interest generated is used to support patients	Ring-fenced budget allocation	Endowment and investments: accrued interest used to finance procurement
Financing source	Government and private donations (3 to 1 government matching of donations)	Government funding (earmarking): pharmaceutical price regulation scheme (PPRS)[64]	Government and donations (various investment strategies)[65]
Governance	Managed by the KKH Health Fund (KKHHF), which is part of the SingHealth Fund and supported by the Ministry of Health	Department of Health, Scotland	Ministry of Health, the Aid and Debt Management Department, Royal Monetary Authority of Bhutan
Scope	Medicines approved by the rare disease fund committee, after other financial schemes have been exhausted	Drugs approved by the Scottish Medicines Consortium (SMC) or on a case-by-case basis	To finance procurement of essential drugs and vaccines in Bhutan
Incentives	Donations are 250% tax deductible	–	Tax-free status

In Malaysia, the Rare Disease Alliance Foundation Malaysia (RDAFM) was established as a trust registered under the Trustees (Incorporation) Act 1952 in September 2017. The foundation comes under the purview of the Prime Minister's Department, and therefore, reports to it. The government has indicated its expectation for the RDAFM to drive and manage the Rare Diseases Trust Fund, also known as Tabung Amanah Penyakit Jarang Jumpa, with support from the Ministry of Health. In terms of funding sources, it was indicated by the government that the trust fund will be sustained by fundraising efforts led by patient groups, which will be insufficient to cover the high cost of treatment, especially on a sustainable basis. While the detailed scope of this trust fund is not yet available, from our roundtable discussion it is understood that the funds allocated aim at covering the cost of treatment and providing additional income support to affected families. Taken altogether, the existing model for the rare disease trust fund in Malaysia poses a few challenges relating to sustainability and governance of the trust fund, as highlighted in IDEAS' previous policy paper (see Table 9).[4]

Table 9: Policy Recommendations for the Rare Disease Trust Fund in Malaysia.

Model	Recommendations
Public finance contribution	• Absorb ring-fenced allocation within the trust fund as the basis for an annual budget OR • Endow the fund with sufficient capital to generate interest
Financial incentives for charitable donations	• As a minimum, ensure donations to the trust fund are tax deductible AND/OR • Introduce match-funding of charitable contributions AND/OR • Introduce more generous tax incentives (e.g. double deductions)
Confidence building for charitable incentives	• Take steps to raise awareness and build confidence in corporate donations for rare diseases (e.g. letters of support)
Scope of fund	• Define clearly the scope of the fund – if only for orphan drugs – then ensure other needs are met in the wider healthcare system
Treatment eligibility	• Define clear eligibility criteria for treatments accessible through the fund, consistent with the wider public healthcare system but recognising the unique challenges posed by rare diseases
Patient eligibility	• Define clear eligibility criteria for patients accessing the scheme, which could include some form of means testing

Governance	• The MOH should oversee the governance of the trust fund and be the ultimate decision maker AND • A decision-making committee representing other stakeholders should be established to support the MOH AND • A committee of technical experts including clinicians should be established to support decision-making

Source: RDF, Bhutan Health Trust Fund, NICE

2.6 Developing the pharmaceutical industry to promote generic competition for drugs treating rare diseases

Generic drugs are important components of measures introduced by healthcare regulatory authorities to reduce treatment costs. Local manufacturing of generic drugs is considered a top priority in the National Medicine Policy 2006 and Malaysia's National Transformation Program 2010. At present, the local manufacturers are capable of producing 80% of the medicines in the Malaysian National Essential Drugs List (NEDL). In fact, Malaysia's generic market share has continued to grow over the years, even faster than innovator drugs. Generic drug sales consisted of 55% of total prescription sales by value, with an increment of 9% over a period of 2 years.[66]

As the patent protection and exclusivity period of some orphan drugs are expiring soon, there is an increasing interest in the manufacturing of generic orphan drugs. However, there are a few prerequisites for the successful generic manufacturing of orphan drugs in Malaysia. Many of the orphan drugs are biopharmaceuticals that are manufactured from living cells and hence may undergo extensive post translational modification including glycosylation and amidation. These post translational modifications, which are highly dependent on production cell lines, as well as growth and purification methods, are highly sensitive to changes in production conditions. Seemingly minor changes in the production process may have a significant impact on the efficacy and safety of the end product. Thus, Malaysia needs to create suitable advanced facilities for industry grade cell line production in the country to ensure successful manufacturing of the drugs.

In most patients and conditions, the switch from a branded drug to its generic counterpart is performed with no major complications. However, evidence from complex diseases suggests that generic substitution requires careful evaluation in some settings, and that current bioequivalence criteria may not always be adequate for establishing the interchangeability of branded and generic products. Biopharmaceutical products such as orphan drugs can induce the formation of antidrug antibodies. While this may be harmless in many cases, in other cases it can reduce the efficacy of the drug and induce severe adverse reactions including anaphylaxis or cross reactions with endogenous proteins. Targets of biopharmaceuticals are often specific for humans and not expressed in test animals, which limits the predictability of pre-clinical safety studies. Hence, additional clinical tests might be required for generic orphan drugs to ensure their safety and efficacy.

PILLAR 3: STRENGTHENING GOVERNANCE FOR HOLISTIC MANAGEMENT OF RARE DISEASES

3.1 Institutionalising a legal framework for rare diseases to protect patients from heightened vulnerability

An assessment of the existing legislative landscape in Malaysia reveals a significant dearth of consideration for rare disease management. In contrast to countries such as the Philippines, where the Rare Diseases Act of the Philippines provides a comprehensive policy to address the management of rare diseases, Malaysia does not have a dedicated law nor an official definition of "rare diseases". Relevant acts, such as the Persons with Disabilities Act 2008 (Akta OKU 2008) or the Sale of Drugs Act 1952 contain no explicit mention of rare diseases, though the latter does include the provision that orphan drugs have to fulfil all the requirements of good manufacturing practice (GMP) and good distribution practice (GDP).[9]

Regardless, it is worth noting that the Ministry of Health has recently published the Malaysian Orphan Medicines Guideline establishing the definition of relevant terminologies and guidelines to facilitate access to orphan medicines. The guideline features a comprehensive list of rare diseases in its appendix, whereby this initiative falls under the Ministry of Health's Rare Disease Programme Strategic Plan for 2022 to 2023 which encompasses preventive, curative, and rehabilitative care as well as registry and research elements.[67]

> ***"To further elevate the progress made thus far on the governance of rare diseases, a dedicated legislation, namely a Rare Disease and Orphan Drugs Act, must be instituted."***

As previously reiterated in our earlier IDEAS publication, the rights of rare disease patients, given their heightened vulnerability, must be protected through the establishment of a legal framework.[4] Taiwan's Rare Disease and Orphan Drug Act 2000 can be employed as a benchmark in doing so—approximately 20 years post-implementation, Taiwan now has 225 diseases recognised as rare diseases and 62 orphan drugs granted market authorisation—proving the impact of the Act in facilitating patient access to orphan drugs.[68] Further discussion on the Orphan Drugs Act can be found in Pillar 2.

3.2 Interministerial coordination and collaboration as key to improving integration and continuity of care

Under the existing National Rare Disease Committee implemented by the Ministry of Health in 2019, the governance leadership spans an extensive horizontal network of ministries including the Ministry of Health (MOH), the Ministry of Women, Family and Community Development (KPWKM), the Islamic Development Department of Malaysia (JAKIM), the Ministry of Higher Education (MOHE) as well as other associations. Given that the plight of rare disease patients does not end at mere healthcare complexities, extending across various other facets of life whether in employment and education, this translates into the need for interministerial coordination to ensure the deliverance of holistic, all-around patient care. The priority is to ensure patients living with rare diseases are able to live comfortably with sufficient access not only to their healthcare, but also social wellbeing. Instead of isolated management of rare disease issues within each ministry, the proposed rare disease legislation can institute an interministerial governance of the issues to allow better coordination and integration.

Patient Sharing #3

Unveiling Vasculitis: Atikah's Struggle with a Delayed Diagnosis

In 2008, Atikah, a 21-year old, woke up with vision loss in her left eye. Initially diagnosed with optic neuritis (the inflammation of an optic nerve) in a specialist hospital in Klang Valley, she received steroid medication as treatment and was not recommended to go for follow-up checkups.

Five years later, disaster struck. Atikah woke up with numbness in her lower limbs, realising that she had urinated in her bed. Rushed to a nearby clinic and subsequently transferred to a teaching hospital's neurology department—and after a *false diagnosis*—Atikah was eventually diagnosed with Antineutrophilic Cytoplasmic Antibody (ANCA)-negative Vasculitis, an autoimmune disease where white blood cells called neutrophils would attack small blood vessels, causing inflammation.

The 5-year diagnostic delay is an example of a major barrier that vasculitis patients undergo: *accurate* and *early* diagnosis.

> ***"Vasculitis is a new disease in Malaysia, an autoimmune disease, and I think most of the patients with Vasculitis only get diagnosed later on in life when they are above 20 years old."***

The health complications that follow an accurate diagnosis also necessitate a range of medications, which inevitably means a heavy financial burden. Atikah's OKU card, initially issued due to her impaired vision, only covered hospital admission expenses like X-rays, MRIs, and blood tests. This meant that all of her out-of-hospital treatment, supplements, and surgery bills had to be borne solely by Atikah's family.

> ***"At the orthopaedics [department], I had to get Ankle Foot Orthoses (AFO), which is a device that prevents stiffness in my feet. So, I have to pay out of pocket for that. I also had to go to the dentist. And if I get an infection, I have to go to a private clinic to treat my infection. Another condition that I have is [a] neurogenic bladder, I cannot hold my urine, which is why I need to buy diapers."***

In 2016, Atikah had her first stroke and needed to receive intravenous immunoglobulin (IVIG), an injection that contains antibodies to fight the inflammatory response of the immune system. With each bottle costing RM500, and having received 10 bottles over 5 days on one occasion, Atikah's father had to bear a whopping bill of between RM20,000 to RM25,000 as the IVIG injections were not covered by the family's insurance or the OKU card.

The necessary Botox injections were an additional financial burden. Every 3 months, 6 months, or a year, Atikah would require two bottles in 2 weeks, which would amount to RM3,000 each time. She also later discovered that her inability to access free IVIG treatment was due to her diagnosis as an ANCA-negative vasculitis patient. Atikah had to transfer from one hospital to another multiple times due to the difference in funding between teaching hospitals and MoH hospitals before she was finally able to access free IVIG medication, albeit temporarily.

Stressed and overwhelmed with the weight of such difficulties, Atikah was forced to withdraw from university in 2016, leaving her feeling frustrated and left behind compared to her peers.

> ***"My life turned to 360. I cannot hang out with my friends. I cannot attend class; I had to study online and get my diploma degree online. I cannot get a good job or my dream job, while my friends graduated and got jobs. I was jealous and I cried. It was hard, even until now. "***

From her initial diagnosis of optic neuritis more than a decade ago until today, Atikah has fought through a multitude of complications, including gastrointestinal (GI) bleeding, haemorrhage, and multiple instances of strokes. If only an accurate initial diagnosis had been provided, Atikah could have had earlier access to crucial treatment, possibly mitigating some of these complications. Perhaps she could have continued her education if the financial burden of continuous hospitalisations during the 5-year delay had been avoided.

Rare disease patients in Malaysia deserve better, and Atikah's story—one which showcases the physical, financial, and emotional challenges that arise from delayed and inaccurate diagnosis—proves the immediate need to alleviate the plight of rare disease patients.

PILLAR 4: ENHANCING SOCIAL PROTECTION SYSTEMS TO PROMOTE RESILIENCE OF INDIVIDUALS WITH RARE DISEASES AND THEIR CAREGIVERS

Based on findings from the focus group and roundtable discussions, it is evident that improving the lives of those living with rare diseases goes beyond healthcare considerations. Due to the nature of their conditions, many rare disease patients and their families require educational needs support, housing adjustments, social care, financial aid, mental health support, and other welfare support. The World Social Protection Report by the International Labour Organisation highlights the critical role of universal social protection schemes in driving economic transformation that tackles structural inequalities. These schemes have proven to be indispensable, especially as a frontline policy response during the COVID-19 pandemic.[69]

Although rare diseases are commonly perceived as a paediatric issue, that is far from the case. Certain rare diseases manifest in adulthood such as Systemic Lupus Erythematosus (SLE), Huntington's disease, Myotrophic Lateral Sclerosis (ALS), Polycystic Kidney Disease (PKD), Ehlers-Danlos Syndrome (EDS), Adult-Onset Still's Disease (AOSD), and Adult Polyglucosan Body Disease (APBD).

Rare diseases affect individuals of all ages, and managing the conditions associated with rare diseases is a lifelong endeavour with different challenges at various stages of a person's life. A young individual diagnosed with a rare disease may encounter difficulties related to accessing essential services like

occupational and speech therapy, leading to potential child development issues. Meanwhile, adult patients may face discrimination in entering the job market. As an estimated 95% of rare disease patients do not have any approved treatment, the challenges of living with rare diseases should be addressed, as the empowerment of patients is also a prerequisite for health.

> ***"Due to the nature of their condition, many rare disease patients and their families require educational needs support, housing adjustments, social care, financial aid, mental health support, and other welfare support."***

The World Health Organisation defines empowerment in healthcare as "a process through which people gain greater control over decisions and actions affecting their health" through tackling access barriers and reshaping the dynamics between communities and institutions.[70] These conditions are often overlooked by the research, medical, and policy-making spheres. For our social protection systems to be truly inclusive, extending to individuals living with rare diseases, the government must identify and address the risks and vulnerabilities across the lifecycle that not only addresses the direct cost of treatment but also the indirect issues.

4.1 Protecting rare disease patients and householders with improved insurance coverage

Despite having a universal public health system, a significant proportion of healthcare costs are borne privately. Malaysia has seen an increase in out-of-pocket (OOP) spending on healthcare from RM22.6 billion in 2020 to RM24.6 billion in 2021, which is equivalent to 31.5% of the total expenditure on health.[71] OOP spending also constitutes the bulk of private healthcare funding, amounting to 74.7% of total health expenditure by private sources of financing. In contrast, only 16.8% of private healthcare financing is from private insurance.

The low level of healthcare expenditure sourced from private insurance reflects the relatively low penetration of private insurance within Malaysia. In the absence of national health insurance, this also indicates that a large majority of the population does not have health insurance coverage. As for individuals with rare diseases, private insurance coverage is further limited due to the commercial disincentive to provide coverage to patients with genetic or pre-existing conditions.

Furthermore, private insurance policies do not provide coverage for orphan drugs partly due to the lack of a centralised rare disease registry. This results in the lack of data needed to undertake actuarial analysis in order to determine the viability of new rare-disease-inclusive products.[5] Meanwhile, current social insurance mechanisms are also insufficient to cover the costs of orphan drugs. SOCSO benefits are tied to employees and their contributions and hence, would not be applicable to a large proportion of rare disease patients who are children.

> ***"As for individuals with rare diseases, private insurance coverage is even more limited due to the commercial disincentive to provide coverage to patients with genetic or pre-existing conditions. Furthermore, private insurance policies do not provide coverage for orphan drugs partly due to the lack of a centralised rare disease registry."***

There are two main policy recommendations to improve private insurance coverage for individuals with rare diseases:

1. **Regulation**

   The government may consider legislation similar to the Genetic Information Nondiscrimination Act (GINA) of 2008 in the United States, an act that aims to prohibit discrimination on genetic information for health insurance and employment. Genetic information refers to information about an individual's genetic tests, the genetic tests of family members, and the manifestation of a disease in family members of the individual. It specifies unlawful practices for employers such as refusing to hire, firing, limiting, segregating, or discriminating against an employee due to their genetic information, requesting or requiring genetic information or family medical history, or disclosing genetic information. It also highlights that health insurers cannot use genetic information to determine eligibility for insurance or to make coverage, underwriting or premium-setting decisions.

2. **Incentives**

   In order to expand the role of insurance in improving coverage for rare diseases, the government **could expand its existing incentives by creating a new level of insurance coverage, categorised as "advanced"—defined as providing coverage for rare diseases—and introduce higher levels of tax relief for such policies.** Alternatively, the government could **share the costs of certain procedures and treatments in order to improve commercial viability.** For instance, the government could agree to split the cost of genetic testing with private insurers.

It is important to note that a regulatory approach is likely to result in increased costs for the consumer, particularly given the relatively small pool of policy holders. However, incentives are likely to be relatively slow in achieving broader coverage. Hence, it is crucial to adopt a gradual approach in developing private insurance coverage, with a balance between regulation and incentives.

Social insurance schemes can also be established to supplement the public healthcare system to address high healthcare costs. One modest step would be to expand the scope of the MySalam scheme to support patients with rare diseases. Established in 2019, MySalam is a free takaful income assistance scheme for individuals in the B40 income group, funded by the MySalam Trust Fund. Recipients of the Bantuan Sara Hidup (BSH) social security programme aged between 18 to 55 years are eligible for the scheme. Under the scheme, on diagnosis of one of 36 critical illnesses eligible recipients will receive a one-time RM8,000 cash payout and RM50 daily hospitalisation income replacement up to RM700 per annum at any government or qualified hospital. There are two main policy recommendations to improve social insurance coverage for individuals with rare disease through the MySalam scheme:

1. To include rare diseases on the list of critical illnesses for patients to be eligible for the scheme.

2. To include children in the scope of the scheme, such that parents receive benefits when their children are diagnosed.

4.2 Improved coverage and assistance for support services and assistive devices

Providing tax relief and incentives as a form of social protection covering the life cycle of individuals living with rare diseases and their caregivers can be a key component in addressing the inequalities associated with managing these conditions. Government policies such as implementing tax relief for secondary treatment like care services, specialised educational support, and occupational and speech therapy can help alleviate their financial burden by reducing their tax obligations to ensure a more equitable economic environment. Acknowledging the many challenges and costs incurred due to their conditions, tax relief on specialised equipment and home modification deductions needed can also help to offset costs associated with their condition.

> ***"Government policies such as implementing tax relief for secondary treatment like care services, specialised educational support, and occupational and speech therapy can help alleviate their financial burden by reducing their tax obligations to ensure a more equitable economic environment."***

Taking an example close to home, Taiwan's National Health Insurance reimburses the expenses of medical coverage for rare diseases. However, the review process takes a long time. Since 2015, the Ministry of Health and Welfare has offered a six-month medication subsidy for additional expenses not covered or under review, with the aim of safeguarding the patient's well-being.

Patients receive medical support and protection through the Rare Disease Control and Orphan Drug Act, while the Physically and Mentally Disabled Citizens Protection Act focuses on socio-economic empowerment in relation to their condition covering areas such as employment, education, and extended care. Both acts include people with rare diseases with the latter act in Article 71 outlining subsidies to residential, day care, assistive devices, home care, and other expenses related to their welfare.

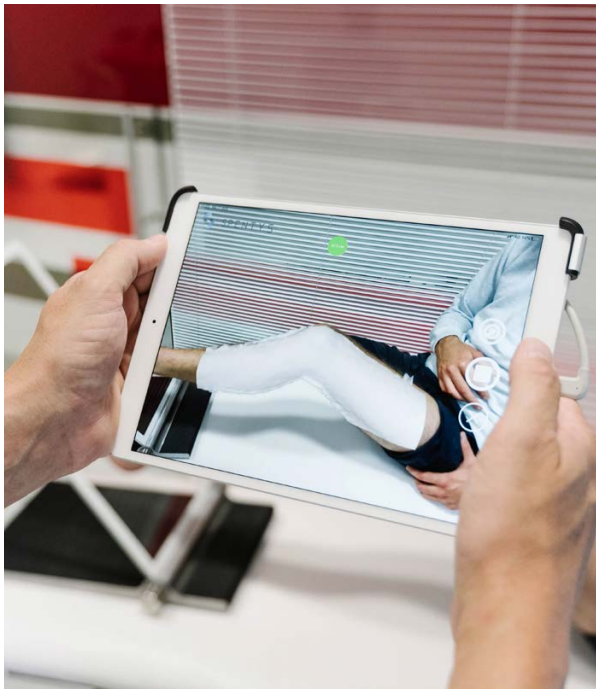
4.3 Promoting inclusive employment opportunities for people living with rare diseases and their caregivers

Social protection of persons with rare diseases in employment can be done through corporate tax incentives that directly encourage the employment of people living with rare diseases, and their caregivers. Some who live with rare diseases have the capacity to work but often are heavily disadvantaged and face discrimination at the workplace.

A study of 140 companies based in the U.S. between 2015-2018 showed that those with disability inclusion efforts have higher revenue, economic profit margins and net income than companies that did not, disproving misconceptions about the cost of disability inclusion.[72] Thus, it is essential for the government to incentivise the employment of individuals with rare diseases and their caregivers. This recognizes the contributions they bring to the labour market while safeguarding their rights to attend to their health-related responsibilities.

Caregivers should be factored in as well. Strategies dealing with care work are key to recognising their vital roles, alongside healthcare institutions, in their support work for people with rare diseases. Policies should be implemented to qualify them for benefits to help in alleviating the cost burden of rare diseases. Care work, which includes caregiving duties for people who live with rare diseases,

and which is often unpaid and unaccounted for, is a key part of our economy in supporting the "wealth-generating" workforce.



Caregivers play a key role in complementing healthcare and social security systems. However they may experience social and economic marginalisation as a result of the heightened time commitment required for their caring responsibilities. In managing the health complications of people living with rare diseases, they make personal sacrifices and shoulder significant financial burdens from the high cost of healthcare treatments, income loss, home modifications and secondary treatment. Thus, they experience challenges in maintaining full time employment due to their responsibilities. Providing dependent tax credits to caregivers responsible for individuals with rare diseases will help in making caregiving more sustainable. Tax incentives should also be developed to encourage hiring caregivers. In turn, caregivers should be allowed to balance their caring responsibilities with paid or unpaid leave, protected by the government. In this way they can support themselves reasonably while supporting the needs of people living with rare diseases.

> ***"Caregivers play a key role in complementing healthcare and social security systems however experience social and economic marginalisation as a result of the heightened time commitment required for their caring responsibilities."***

While Malaysia does provide social support to people with disabilities, the benefits should extend to individuals living with rare diseases and their caregivers. OKU cardholder benefits include double tax relief for employers who train and hire disabled workers, and fees exemption and company registration renewal for disabled entrepreneurs. Tax relief for taxpayers who have children or spouses with disabilities and for the purchase of special equipment related to disabilities is also provided to OKU cardholders. As some individuals living with rare diseases do have disabling aspects that are not immediately visible with their symptoms fluctuating from time to time, they are unable to qualify for the OKU card and its benefits.

Extending the OKU cardholder benefits to people living with rare diseases and their caregivers, regardless of whether they suffer from functional impairment or not, recognizes their challenges. Providing healthcare equity and social protection will enable them to be active and productive members of society.

Conclusion

Patients living with rare diseases and their caregivers face disproportionate challenges navigating the patient pathway of the current Malaysian healthcare system. At the same time, they are confronted by societal discrimination and inequities in mainstream employment and education systems. From obtaining a diagnosis to accessing treatments to manage their condition, the patient pathway for persons living with rare diseases is an uphill battle against the limited availability of knowledge, training, diagnostics, and treatment resources needed to obtain a precise diagnosis and treatment of the patient's condition.

Besides suffering from the health consequences of their condition alongside the despair and confusion that come with the limited treatment options and support available, patients and their caregivers often have to bear high financial costs. To maintain the status quo with regard to our existing rare disease policy would mean that rare disease patients in Malaysia will continue to be heavily reliant upon ad-hoc government budget allocation and crowdfunding to finance expensive treatments. This is not guaranteed, nor sustainable.

With that, it is clear that Malaysia is in dire need of a more sustainable and innovative financing model that balances the promotion of ongoing innovation and drug development for rare diseases—particularly those without available treatments at present—with cost containment of innovative treatments to ensure equitable access.

As such, policymakers must consider the potential benefits and risks associated with policy reforms that would improve evidence generation and increase incentives for orphan drugs. The pricing of orphan drugs should be regulated directly or indirectly to improve affordability, while not undermining future investments and innovation. A key enabler to finding that balance is through establishing strong and symbiotic public-private partnerships. Meanwhile, recognising that a significant proportion of individuals living with rare diseases do not have any available treatment, the role of trust funds and insurance must also be strengthened to address the unmet needs of such patients.

The impact of living with rare diseases goes beyond health and permeates every aspect of the patients' lives; patients have to face societal barriers that further exclude them and their caregivers from social and economic life. Managing rare diseases entails high expenses and is often coupled with low income, putting rare disease patients at risk of financial hardship. Therefore, there is a pressing need for a sustainable and holistic healthcare and support system that empowers rare disease patients and their caregivers. This system should encompass effective governance and holistic management of rare diseases that ensures affordable access to diagnosis and treatment in our healthcare system, whilst also integrating social protection mechanisms. Rare disease patients and their caregivers can lead productive and dignified lives, as long as a national rare disease strategy that is patient-centric and equitable is put into place.

References

1 Rare Diseases International (RDI). UN Resolution on persons living with a rare disease. 2021. [Online]. Available from: https://www.rarediseasesinternational.org/wp-content/uploads/2022/01/Final-UN-Text-UN-Resolution-on-Persons-Living-with-a-Rare-Disease-and-their-Families.pdf [Accessed 30 August 2023].

2 World Economic Forum. Global data access for solving rare disease—a health economics value framework. 2020. [Online]. Available from: https://www3.weforum.org/docs/WEF_Global_Data_Access_for_Solving_Rare_Disease_Report_2020.pdf

3 Haendel M, Vasilevsky N, Unni D, Bologa C, Harris N, Rehm H, Hamosh A, Baynam G, Groza T, McMurry J, Dawkins H. How many rare diseases are there? Nature Reviews Drug Discovery. 2020 Feb;19(2):77-78. [Online]. Available from: https://doi.org/10.1038/d41573-019-00180-y

4 Todd L, Rao V. Next steps for rare diseases in Malaysia: improving access to treatments [Online]. IDEAS. 2020. Available from: https://www.ideas.org.my/publications-item/policy-paper-no-67-next-steps-for-rare-diseases-in-malaysia-improving-access-to-treatments/

5 Thong MK, Ahmad-Annuar A, Todd L, Mogan Rao V. Whitepaper: rare diseases in Malaysia [Online]. IDEAS. 2019. Available from: https://www.ideas.org.my/publications-item/whitepaper-rare-diseases-in-malaysia/

6 Shafie AA. Policy IDEAS No 58 Improving access to orphan drugs in Malaysia [Online]. IDEAS. 2019. Available from: https://www.ideas.org.my/publications-item/policy-ideas-no-58-improving-access-to-orphan-drugs-in-malaysia/

7 Thong MK, Ahmad-Annuar A. National policy on rare diseases living with dignity: in search of solutions for rare diseases [Online]. IDEAS. 2018. Available from: https://www.ideas.org.my/publications-item/brief-ideas-no-10-national-policy-on-rare-diseases-living-with-dignity-in-search-of-solutions-for-rare-diseases/

8 Osio-Salido E, Manapat-Reyes H. Epidemiology of Systemic Lupus Erythematosus in Asia. Lupus. 2010 Oct;19(12):1365-1373. [Online]. Available from: https://doi.org/10.1177/0961203310374305

9 Shafie AA, Supian A, Ahmad Hassali MA, Ngu LH, Thong MK, Ayob H, Chaiyakunapruk N. Rare disease in Malaysia: challenges and solutions. PLoS One. 2020 Apr 02;15(4):e0230850. [Online]. Available from: https://doi.org/10.1371/journal.pone.0230850

10 Levesque JF, Harris MF, Russell G. Patient-centred access to health care: conceptualising access at the interface of health systems and populations. International Journal for Equity in Health. 2013 Dec;12:1-9. [Online]. Available from: https://doi.org/10.1186/1475-9276-12-18

11 Yan X, He S, Dong D. Determining how far an adult rare disease patient needs to travel for a definitive diagnosis: a cross-sectional examination of the 2018 national rare disease survey in China. International Journal of Environmental Research and Public Health. 2020 Mar;17(5):1757. [Online]. Available from: https://doi.org/10.3390/ijerph17051757

12 Accurate diagnosis of rare diseases remains difficult despite strong physician interest—Global Genes. Global Genes. 2014. [Internet]. Available from: https://globalgenes.org/blog/accurate-diagnosis-of-rare-diseases-remains-difficult-despite-strong-physician-interest-2/

13 National Health and Morbidity Survey 2019. Non-communicable diseases, healthcare demand, and health literacy: key findings. [Online]. Available from: https://iptk.moh.gov.my/images/technical_report/2020/4_Infographic_Booklet_NHMS_2019_-_English.pdf

14 Katavic SS, Tanackovic SF, Badurina B. Illness perception and information behaviour of patients with rare chronic diseases. Information Research: An International Electronic Journal. 2016 Mar;21(1):n1. [Online]. Available from: https://files.eric.ed.gov/fulltext/EJ1094564.pdf

[15] Iriart JA, Nucci MF, Muniz TP, Viana GB, Aureliano WD, Gibbon S. From the search for diagnosis to treatment uncertainties: challenges of care for rare genetic diseases in Brazil. Ciência & Saúde Coletiva. 2019 Sep 26;24:3637-50. [Online]. Available from: https://www.scielo.br/j/csc/a/vgJYDtBJhpBBzj44Sz76btG/?format=html&lang=en

[16] Liljesthröm, M.; Armando, R.; Maier, B.; Robert, F.; Zelikman, D. Rare diseases in Argentina: what a survey of families tell us about the diagnosis process. 2015. [Online]. Available from: https://doi.org/10.3390%2Fijerph20064732

[17] Shafie AA, Chaiyakunapruk N, Supian A, Lim J, Zafra M, Hassali MA. State of rare disease management in Southeast Asia. Orphanet Journal of Rare Diseases. 2016 Aug 02;11(107):1-11. [Online]. Available from: https://doi.org/10.1186/s13023-016-0460-9

[18] Malaysian Medical Council (MMC). Medical education & recognition. [Internet]. Available from: https://mmc.gov.my/medical-education-recognition

[19] Kementerian Kesihatan. Senarai hospital kerajaan [List of government hospitals in Malaysia]. Kementerian Kesihatan. [Internet]. Available from: https://www.moh.gov.my/index.php/database_stores/store_view/82

[20] Genetic Counselling Society of Malaysia. Genetic services and training. [Internet]. Available from: https://gcsocietymalaysia.org.my/genetic-services-and-training/

[21] CodeBlue. Genetic counselling under MOH only available in HKL, Penang Hospital. 27 October 2021. [Internet]. Available from: https://codeblue.galencentre.org/2021/10/27/genetic-counselling-under-moh-only-available-in-hkl-penang-hospital/

[22] Ramsey-Goldman R, Schilling EM, Dunlop D, Langman C, Greenland P, Thomas RJ, Chang RW. A pilot study on the effects of exercise in patients with Systemic Lupus Erythematosus. Arthritis Care & Research. 2001 May 15;13(5):262-269. [Online]. Available from: https://doi.org/10.1002/1529-0131(200010)13:5%3C262::aid-anr4%3E3.0.co;2-8

[23] van den Berg L, Favejee M, Wens S, Kruijshaar M, Karstens F, Rozenberg R, Praet S, Reuser A, Pijnappel P, Bussmann J, van Doorn P. Effects of exercise training in 23 adults with Pompe disease receiving enzyme therapy. BMC Musculoskeletal Disorders. 2013 May 29;14(Suppl 2):P17. [Online]. Available from: https://doi.org/10.1186%2F1471-2474-14-S2-P17

[24] Plaiasu V, Nanu M, Matei D. Rare disease day—at a glance. Maedica. 2010 Jan;5(1):65. [Online]. Available from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3150084/

[25] CodeBlue. MOH Forming Trust Fund, Database for rare diseases. 16 March 2022. [Internet]. Available from: https://codeblue.galencentre.org/2022/03/16/moh-forming-trust-fund-database-for-rare-diseases/

[26] Owings, L. What happens to people in South Africa who have rare diseases? 06 Sep 2021. [Online]. Available from: https://www.dailymaverick.co.za/article/2021-09-06-what-happens-to-people-in-south-africa-who-have-rare-diseases

[27] Deif, I. Marking rare disease day in Egypt: doctors weigh challenges, highlight breakthroughs. 15 Mar 2017. [Online]. Available from: https://english.ahram.org.eg/NewsContent/7/48/260974/Life—Style/Health/Marking-Rare-Disease-Day-in-Egypt-Doctors-weigh-ch.aspx

[28] Adachi T, El-Hattab AW, Jain R, Nogales Crespo KA, Quirland Lazo CI, Scarpa M, Summar M, Wattanasirichaigoon D. 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. 2023 Mar 08;20(6):4732. [Online]. Available from: https://doi.org/10.3390/ijerph20064732

[29] Nothaft W, Goldsmith C, Le Cam Y. It takes far too long for a rare disease to be diagnosed. In: World Economic Forum 2020 Feb 28 (Vol. 302). [Online]. Available from: https://www.weforum.org/agenda/2020/02/it-takes-far-too-long-for-a-rare-disease-to-be-diagnosed-heres-how-that-can-change/

[30] Noraihan MN, See MH, Raja R, Baskaran TP, Symonds EM. Audit of birth defects in 34,109 deliveries in a tertiary referral centre. The Medical Journal of Malaysia. 2005 Oct;60(4):460-8.

[31] Leong YH, Gan CY, Tan MA, Majid MI. Present status and future concerns of expanded newborn screening in Malaysia: sustainability, challenges and perspectives. The Malaysian Journal of Medical Sciences: MJMS. 2014 Mar;21(2):63.

[32] Dharssi S, Wong-Rieger D, Harold M, Terry S. Review of 11 national policies for rare diseases in the context of key patient needs. Orphanet Journal of Rare Diseases. 2017 March 31;12(63):1-3. [Online]. Available from: https://ojrd.biomedcentral.com/articles/10.1186/s13023-017-0618-0?report=reader

[33] Lewin Group for the Evelyn Foundation. The national economic burden of rare disease study. 2021. [Online]. Available from: https://everylifefoundation.org/wp-content/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf

[34] NGO Committee for Rare Diseases. The "right to health" in rare diseases: a practical contribution to implementing and achieving the Sustainable Development Goals. 2018. [Online]. Available from: https://www.ngocommitteerarediseases.org/wp-content/uploads/2018/05/NGO-CfRDs-Submission-The-Right-to-Health-in-Rare-Diseases_Feb-15-2018.pdf

[35] Black N, Martineau F, Manacorda T. Diagnostic odyssey for rare diseases: exploration of potential indicators. Policy Innovation Research Unit (PIRU). 2015. [Online]. Available from: https://piru.ac.uk/assets/files/Rare%20diseases%20Final%20report.pdf

[36] Ninomiya K, Okura M. Nationwide comprehensive epidemiological study of rare diseases in Japan using a health insurance claims database. Orphanet Journal of Rare Diseases. 2022 Dec;17(1):1-3. [Online]. Available from: https://ojrd.biomedcentral.com/articles/10.1186/s13023-022-02290-0

[37] Kaufmann P, Pariser AR, Austin C. From scientific discovery to treatments for rare diseases–the view from the National Center for Advancing Translational Sciences–Office of Rare Diseases Research. Orphanet Journal of Rare Diseases. 2018 Dec;13:1-8. [Online]. Available from: https://doi.org/10.1186/s13023-018-0936-x

[38] Aronson JK. Rare diseases and orphan drugs. British Journal of Clinical Pharmacology 2006 Mar 1;61(3):243–5. [Online]. Available from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1885017/

[39] Chambers JD, Silver MC, Berklein FC, Cohen JT, Neumann PJ. Orphan drugs offer larger health gains but less favourable cost-effectiveness than non-orphan drugs. Journal of General Internal Medicine. 2020 Sep;35:2629-36. [Online]. Available from: https://doi.org/10.1007/s11606-020-05805-2

[40] Abu Bakar F, Shaharir SS, Mohd R, Kamaruzaman L, Mohamed Said MS. Work disability in a multi-ethnic Malaysian Systemic Lupus Erythematosus cohort: a cross-sectional study. International journal of rheumatic diseases. 2019 Jun;22(6):1002-7. [Online]. Available from: https://doi.org/10.1111/1756-185x.13572

[41] Czech M, Baran-Kooiker A, Atikeler K, Demirtshyan M, Gaitova K, Holownia-Voloskova M, Turcu-Stiolica A, Kooiker C, Piniazhko O, Konstandyan N, Zalis' ka O. A review of rare disease policies and orphan drug reimbursement systems in 12 Eurasian countries. Frontiers in Public Health. 2020 Jan 28;7:416. [Online]. Available from: https://doi.org/10.3389/fpubh.2019.00416

[42] Dong D, Wang Y. Challenges of rare diseases in China. The Lancet. 2016 May 7;387(10031):1906. [Online]. Available from: https://doi.org/10.1016/S0140-6736(16)30418-4

[43] Koçkaya G, Atalay S, Oğuzhan G, Kurnaz M, Ökçün S, Sar Gedik Ç, Şaylan M, Şencan N. Analysis of patient access to orphan drugs in Turkey. Orphanet Journal of Rare Diseases. 2021 Dec;16:1-8. [Online]. Available from: https://doi.org/10.21203/rs.3.rs-93956/v1

[44] Noonan Syndrome Awareness Association. The challenges of parenting a child with a rare disease. 2017. [Internet]. Available from: https://noonansyndrome.com.au/the-challenges-of-parenting-a-child-with-a-rare-disease

[45] CORD. Canada's rare disease caregivers under immense stress, struggling with mental health issues, isolation and financial burden. [Online]. Available from: https://www.raredisorders.ca/content/uploads/CORD_NationalFamilyCaregiverDay_PressRelease_04022019_FINAL1.pdf

[46] Daly A. Impact of rare disease quality of life: what it means in real terms for PLWRD (person living with a rare disease). 2018. [Online]. Available from: http://download2.eurordis.org.s3.amazonaws.com/ecrd/ECRD_2018/Speaker_Presentations/0401-Daly.pdf

[47] Bogart KR, Irvin VL. Health-related quality of life among adults with diverse rare disorders. Orphanet journal of rare diseases. 2017 Dec;12:1-9. [Online]. Available from: https://doi.org/10.1186/s13023-017-0730-1

[48] Reinhard C., Bachoud-Lévi A., Bäumer T., Bertini E., Brunelle A., Buizer A. et al.. The European reference network for rare neurological diseases. Frontiers in Neurology 2021;11. [Online]. Available from: https://doi.org/10.3389/fneur.2020.616569

[49] Aitken M, Kleinrock M, Muñoz E, Porwal U. Orphan drugs in the United States: rare disease innovation and cost trends through 2019. IQVIA. [Online]. Available from: https://www.iqvia.com/insights/the-iqvia-institute/reports/orphan-drugs-in-the-united-states-rare-disease-innovation-and-cost-trends-through-2019

[50] Ministry of Health Malaysia. Malaysian Orphan Medicines Guideline 2020. [Internet]. Available from: https://www.pharmacy.gov.my/v2/en/documents/malaysian-orphan-medicines-guideline-2020.html

[51] National Pharmaceutical Regulatory Agency (NPRA). Designation and registration of orphan medicines. [Online]. Available from: https://www.npra.gov.my/easyarticles/images/users/1047/drgd/APPENDIX-13—Designation-and-Registration-of-Orphan-Medicines.pdf

[52] Pearson C, Schapiro L, Pearson SD. The next generation of rare disease drug policy: ensuring both innovation and affordability. Journal of Comparative Effectiveness Research. 2022 Oct;11(14):999-1010. [Online]. Available from: https://icer.org/wp-content/uploads/2022/04/ICER-White-Paper_The-Next-Generation-of-Rare-Disease-Drug-Policy_040722.pdf

[53] Lim Y, Lajis R, Mohd Din R, Md Yusof FA. A preliminary approach to patient access scheme in Malaysia: current experience and future direction. Value in Health. 2018 Sep; 21:S112. [Online]. Available from: https://doi.org/10.1016/j.jval.2018.07.855

[54] Klemp M, Frønsdal KB, Facey K. What principles should govern the use of managed entry agreements? International Journal of Technology Assessment in Health Care. 2011 Jan; 27(1): 77–83. [Online]. Available from: https://doi.org/10.1017/S0266462310001297

[55] Ashraf A, Ong S. Improving patient access scheme in Malaysia: lessons from Italy. International Journal of Pharmaceutical and Healthcare Marketing. 2023;17(3):327-339. [Online]. Available from: https://doi.org/10.1108/ijphm-06-2022-0059

[56] Ibrahim S, Muniandy G, Wan Ibrahim WNA, Khalil F, Yahya NF. Impact of medicines pooled procurement towards medicines pricing in the public sector. Proceedings of the 11th HTAsiaLink Annual Conference; 2023; Putrajaya, Malaysia.

[57] Teerawattananon Y, Rattanavipapong W, Lin L, Dabak S, Gibbons B, Isaranuwatchai W et al. Landscape analysis of Health Technology Assessment (HTA): systems and practices in Asia. International Journal of Technology Assessment in Healthcare 2019;35(6):416-421. [Online]. Available from: https://doi.org/10.1017/s0266462319000667

[58] The ASEAN Secretariat. ASEAN follow-up assessment on the current status of Health Technology Assessment (HTA) institutionalisation and capacity needs of ASEAN Member States. [Online]. Jakarta; 2021. Available from: https://asean.org/wp-content/uploads/2021/09/ASEAN-FUA-Final-Report-FINAL-2021.09.09.pdf

[59] Beneluxa Initiative. Health Technology Assessment [Internet]. Vienna (AT): Beneluxa [updated 2021; cited 2023 Sep 1]. Available from: https://beneluxa.org/hta

[60] Sharma M, Teerawattananon Y, Dabak SV, Isaranuwatchai W, Pearce F, Pilasant S, Sabirin J, Mayxay M, Guerrero M, Phuong NK, Sastroasmoro S. A landscape analysis of Health Technology Assessment capacity in the Association of South-East Asian Nations region. Health Research Policy and Systems. 2021 Dec;19:1-3. [Online]. Available from: https://doi.org/10.1186/s12961-020-00647-0

[61] APEC Rare Disease Network: Addressing barriers to the diagnosis and treatment of rare disease in APEC. [Internet]. Available from: https://www.apec.org/rarediseases

[62] Rare Disorders Society Singapore. Rare Disorders Society (Singapore)—we carry hope [Internet]. 2020. Available from: https://www.rdss.org.sg/

[63] SWFI Institute. Bhutan Health Trust Fund. [Internet]. 2023. [cited 2023 August 17]. Available from: https://www.swfinstitute.org/profile/61e8d855c7a246872951e8f7

[64] GOV.UK. Pharmaceutical price regulation scheme 2014 [Internet]. GOV.UK. Available from: https://www.gov.uk/government/publications/pharmaceutical-price-regulation-scheme-2014

[65] Ministry of Health. Policy brief of healthcare financing in Bhutan (2018-2020): evidence from the National Health Accounts. May 2021. [Online]. Available from: https://www.moh.gov.bt/wp-content/uploads/ict-files/2021/07/Policy-Brief-on-the-National-Health-Accounts-2021.pdf

[66] Malaysia Competition Commission (MyCC). Market review on priority sector under Competition Act 2010 – Pharmaceutical Sector [Internet]. 2017 Dec. Available from: https://www.mycc.gov.my/sites/default/files/pdf/newsroom/Market%20Review%20on%20Pharmaceutical%20Sector.pdf

[67] CodeBlue. MOH allocated RM10Mil for rare disease treatment in 2021. [Internet]. 2021 [cited 2023 August 28]. Available from: https://codeblue.galencentre.org/2021/10/18/moh-allocated-rm10mil-for-rare-disease-treatment-in-2021

[68] Hsiang NC, Huang WF, Gau CS, Tsai TW, Chang LC. The impact of the rare disease and Orphan Drug Act in Taiwan. Journal of Food and Drug Analysis. 2021 Dec 14;29(4):717–25. [Online]. Available from: https://doi.org/10.38212/2224-6614.3383

[69] World Social Protection Report 2020–22: Social protection at the crossroads—in pursuit of a better future, Geneva: International Labour Office, 2021. [Online]. Available from: https://www.ilo.org/wcmsp5/groups/public/@ed_protect/@soc_sec/documents/publication/wcms_817572.pdf

[70] Health promotion glossary of terms 2021. Geneva: World Health Organisation; 2021. Licence: CC BY-NC-SA 3.0IGO. [Online]. Available from: https://www.who.int/publications/i/item/9789240038349

[71] CodeBlue. Report: Malaysia's out-of-pocket expense rises with higher spending on drugs, inpatient services [Internet]. 2023. Available from: https://codeblue.galencentre.org/2023/02/24/report-malaysias-out-of-pocket-expense-rises-with-higher-spending-on-drugs-inpatient-services/

[72] Accenture, Disability: IN, The American Association of People with Disabilities. Getting to equal: the disability inclusion advantage. 2018. [Online]. Available from: https://www.accenture.com/_acnmedia/PDF-89/Accenture-Disability-Inclusion-Research-Report.pdf

IDEAS
INSTITUTE FOR DEMOCRACY AND ECONOMIC AFFAIRS

The Institute for Democracy and Economic Affairs (IDEAS) is a nonprofit research institute based in Malaysia dedicated to promoting solutions to public policy challenges. Our vision is :

"A Malaysia that upholds the principles of liberty and justice"

Our mission at IDEAS is to improve the level of understanding and acceptance of public policies based on the principles of rule of law, limited government, competitive markets and free individuals. Our work is independent of vested interests and partisan influences. We have also expanded our work into new areas focussing on our three overarching missions – advancing a competitive economy, ensuring trust in institutions and promoting an inclusive Malaysia. We act as an intellectual centre creating space for cross partisan principles-centric and results-oriented dialogue.

We achieve this by:

- Publishing cutting-edge research
- Initiating dialogues with government, lawmakers, businesses and civil society
- Providing thought leadership
- Facilitating networking between like-minded individuals
- Organising educational programmes

Please support us by making a donation. You can make a contribution by cheque payable to "IDEAS Policy Research Berhad" or by transfer to our account CIMB 8008852042. We can only survive with your support.

IDEAS Policy Research Berhad
The Lower Penthouse
Wisma Hang Sam, 1, Jalan Hang Lekir 50000 Kuala Lumpur

www.ideas.org.my
Reg №: 1219187-V

Selection of IDEAS' Publications (2022-2023)

Policy Ideas

Policy Paper No 77 – Pantau Laksana: Assessing Malaysia's Transparency and Accountability Initiatives in Implementing Emergency Fiscal Responses by Sri Murniati Yusuf, Alissa Rode and Zhen Ting Low (February 2023)

Policy Paper No 76 – Breast Cancer Care in Malaysia: Access to Early Detection, Diagnosis and Optimal Treatment by Fatiha Hana Shabaruddin and Kirjane Ngu (February 2023)

Policy Paper No 75 –The New Economic Policy and Contesting Bumiputera Identity Among Orang Asli and the Indigenous Peoples of Sabah and Sarawak by Wan Zawawi Ibrahim (November 2021)

Brief Ideas

Brief IDEAS No. 38 – Belt and Road Initiative (BRI) Projects and Information Asymmetry: Engagements with Local Communities in Malaysia by Benjamin YH Loh, Imran Shamsunahar and Jazreen Harith (June 2023)

Brief IDEAS No. 37 – Challenges in Hiring and Talent Upskilling in Malaysia as an Upper Middle Income Country: Paving the Way Forward for a Resilient, Skilled Workforce in the Manufacturing and Services Sectors by Imran Shamsunahar, Jazreen Harith and Juita Mohamad (February 2023)

Brief IDEAS No. 36 – Strengthening the Digital Trade Ecosystem: The Next Frontier for Malaysia by Farlina Said, Imran Shamsunahar and Juita Mohamad (December 2022)

Versi Bahasa Malaysi – Polisi Ringkas No. 35 – Ketelusan Belanjawan Di Negeri-negeri Malaysia: Dapatan Utama Indeks Belanjawan Terbuka Malaysia - MYOBI 2022 ditulis oleh Sri Murniati Yusuf, Alissa Rode dan Muhammad Arieff Najmuddin (April 2023)

Brief IDEAS No. 35 – Budget Transparency in Malaysian States: Key Findings of Malaysia's Open Budget Index (MyOBI) 2022 by Sri Murniati Yusuf, Alissa Marianne Rode, and Muhammad Arieff Najmuddin Mohd Mohtar (July 2022)

Brief IDEAS No. 34 – Political financing in Malaysia: Aligning reforms with voters' expectations by Aira Azhar (October 2021)

Report

API Report No.07 – ASEAN Integration Report 2022 by Dr. Evelyn S. Devadason, Dr. Lurong Chen, Ms Yuanita Suhud, Dr. Aya Ono, Dr. Anh Tuan Nguyen, Dr. Poppy S. Winanti, Dr. Katrina Navallo, Dr. Upalat Korwatanasakul, Dr. Adiasri Putri Purbantina, Mr Imran Shamsunahar, Dr. Juita Mohamad, Ms Julia Merican, Ms Kirjane Ngu and Mr Jazreen Harith (November 2022)

Left Far Behind: The Impact of COVID-19 on Access to Education and Healthcare for Refugee and Asylum-Seeking Children in Peninsular Malaysia by Diode Consultancy and Wan Ya Shin (September 2022)

Policy IDEAS are IDEAS' regular publications that introduce and propose ideas for policy reforms based on analysis of existing policies or best practices.

IDEAS
INSTITUTE FOR DEMOCRACY AND ECONOMIC AFFAIRS

Institute for Democracy and Economic Affairs (IDEAS)
The Lower Penthouse. Wisma Hang Sam, 1, Jalan Hang Lekir 50000 Kuala Lumpur

Website: ideas.org.my Tel: +603 2070 8881 / 8882 Fax: +603 2070 8883