White Paper

Rare Diseases in APAC: The Unmet Potential

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Introduction

In our two-part report, IQVIA Asia-Pacific presents an overview of the rare disease landscape in the Asia-Pacific (APAC) region. In *Part 1 – Rare Diseases in APAC: The Unmet Potential*, we examine the burden of rare disease in the APAC region, the level of maturity regarding rare disease care delivery in various APAC markets, and the potential size of these markets. In *Part 2: Unlocking Solutions for Rare Diseases in APAC*, we look at current challenges in rare disease management, potential solutions, and innovations in the rare disease space.

We consider a broad set of countries in the APAC region for our analysis (*Figure 1*) that can be clustered into groups based on macroeconomic indicators such as Gross Domestic Product per capita and healthcare spend per capita.

Globally, the focus on rare diseases has grown considerably over the last few years. Even in the APAC region, this market is gaining momentum owing to the pronounced disease burden and unique challenges that patients face. Countries in the region have varying degrees of maturity regarding rare disease management, with developed markets having more comprehensive ecosystems for care delivery and middle-income countries (e.g. India) still in the midst of establishing comprehensive policies, strengthening their regulatory frameworks to improve access to rare disease medications, and, thus, on the cusp of unlocking significant potential.

Figure 1: set of APAC countries considered in our analysis

Source: IQVIA analysis

Rare disease overview

What is the burden of rare diseases?

"Rare diseases", by definition, are conditions affecting a smaller percentage of people compared with the general population, making it challenging for national healthcare systems to appropriately address care delivery needs and adequately allocate resources to tackle them. Although approximately ~7,000–8,000 rare diseases have been identified, only an estimated ~5% currently have treatments.

Although rare by definition, as a group, rare diseases affect ~6%–8% of the population globally, with World Economic Forum estimates indicating that approximately 350 Mn to 475 Mn people were affected by rare diseases in 2020. Furthermore, over ~80% are genetic in origin and mostly affect children or young adults, with the potential to affect multiple siblings in a family. Unfortunately, with treatments only identified for ~5% of rare diseases, most turn out to be fatal, and reports suggest that almost one-third of children born with rare diseases die before the age of five.

The Asia-Pacific (APAC) region, which is home to more than half (~60%) of the global population, is characterised by extreme diversity in terms of languages, cultures, and socioeconomic backgrounds. It is in this region that the highest number of persons living with rare disease (PLWRD) reside, with estimates indicating more than 258 million individuals are impacted. The region's heterogeneity, coupled with complex healthcare and policy environments, presents unique challenges in the management of rare diseases.

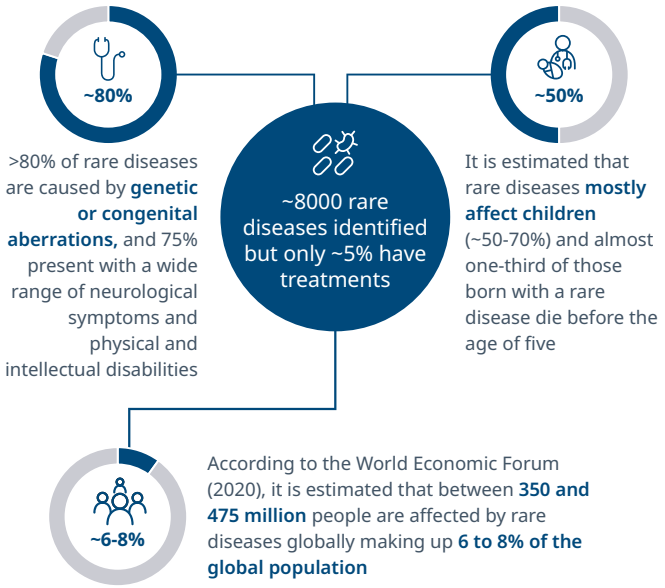
How are rare diseases defined?

Currently, there is no universally accepted and employed definition of rare diseases; thus, multiple definitions exist globally, varying by country. A systematic global study found that there are about ~300 definitions from over a thousand organisations that are largely derived from prevalence thresholds. These definitions are expressed as a fraction, percentage, or rate, i.e. the number of cases per 100,000 population.

Figure 2: Rare disease at a glance

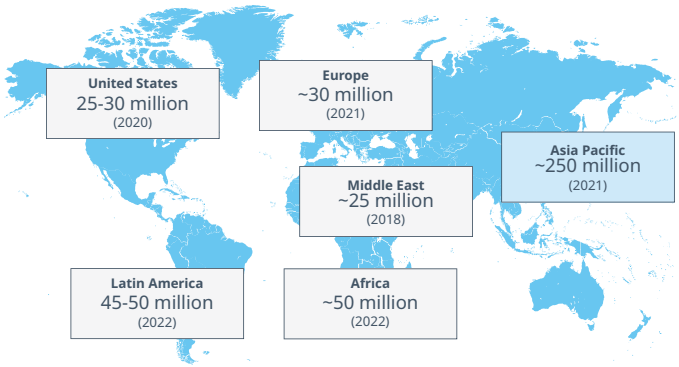
Though ~8,000 rare diseases have been identified, merely an estimated 5% currently have treatments...

Rare Disease Burden (1 of 2)



...and the APAC region, home to 60% of the global population, has the highest number of Persons Living with a Rare Disease

Rare Disease Burden (1 of 2)



Source: IQVIA analysis, Secondary Market Research [3, 4, 8, 10]

Figure 3: the many definitions of rare disease

As the name suggests, rare diseases affect very few people compared to common disorders, although, no universal definition exists

Rare Disease Definition

- The definition of rare disease varies across the world, wherein as per a systematic global review, there are about 296 definitions from 1109 organizations, most of which are explicitly or implicitly derived from prevalence thresholds
- National definitions are critical since this helps design policy measures and determine extent of government support to manage rare diseases

Prevalence

- Disease is considered rare if the prevalence is below a pre-defined threshold (largely expressed as a rate)

Europe
Affects <1 in 2,000 people

Singapore
Affects <1 in 2,000 people

Japan: Affects <1 in 2,000 people

Patient number

- Disease is considered rare if the number of people it affects at a given time is below a pre-defined threshold

United States
Affects <200k people at any given time

Additional dimensions

- Disease is considered rare if it meets pre-defined thresholds of prevalence or patient numbers AND additional criteria (socioeconomic impact, scarcity of treatment options, etc.)

Thailand
Affects < 10,000 people per year AND is severe, associated with difficulties in accessing adequate diagnosis, screening, treatment; and has high economic burden on the family and society

Source: IQVIA analysis, Secondary Market Research [5, 7, 10, 11, 12]

A few countries also consider additional criteria that are relevant from a healthcare system and policy perspective, capturing elements of socioeconomic impact. This is applicable to Thailand, where the Universal Coverage Scheme (UCS) employs a rare disease definition encompassing a combination of the number of cases per population and a broader set of parameters such as severity, diagnosis, treatment, and economic burden (*see Figure 3*).

Such discrepancies in defining rare diseases make it challenging to draw meaningful comparisons across countries in the APAC region and estimate the true disease burden.

Rare Disease Management in APAC

How mature is the APAC rare disease market?

An earlier IQVIA white paper ("Catalyzing Sustainable Funding for Rare Diseases") offered a current snapshot of the rare disease landscape in low- and middle-income countries to analyse public-sector maturity in selected markets for improving access to rare disease treatment. We expand on this research to target countries in the APAC region to understand advancements in rare disease management based on specific criteria: the presence of a rare disease definition and policy, the presence of a ring-fenced fund, an accelerated drug registration pathway for orphan drugs, the presence of centres of excellence, and the implementation of national newborn screening programmes (summarised in *Figure 4*).

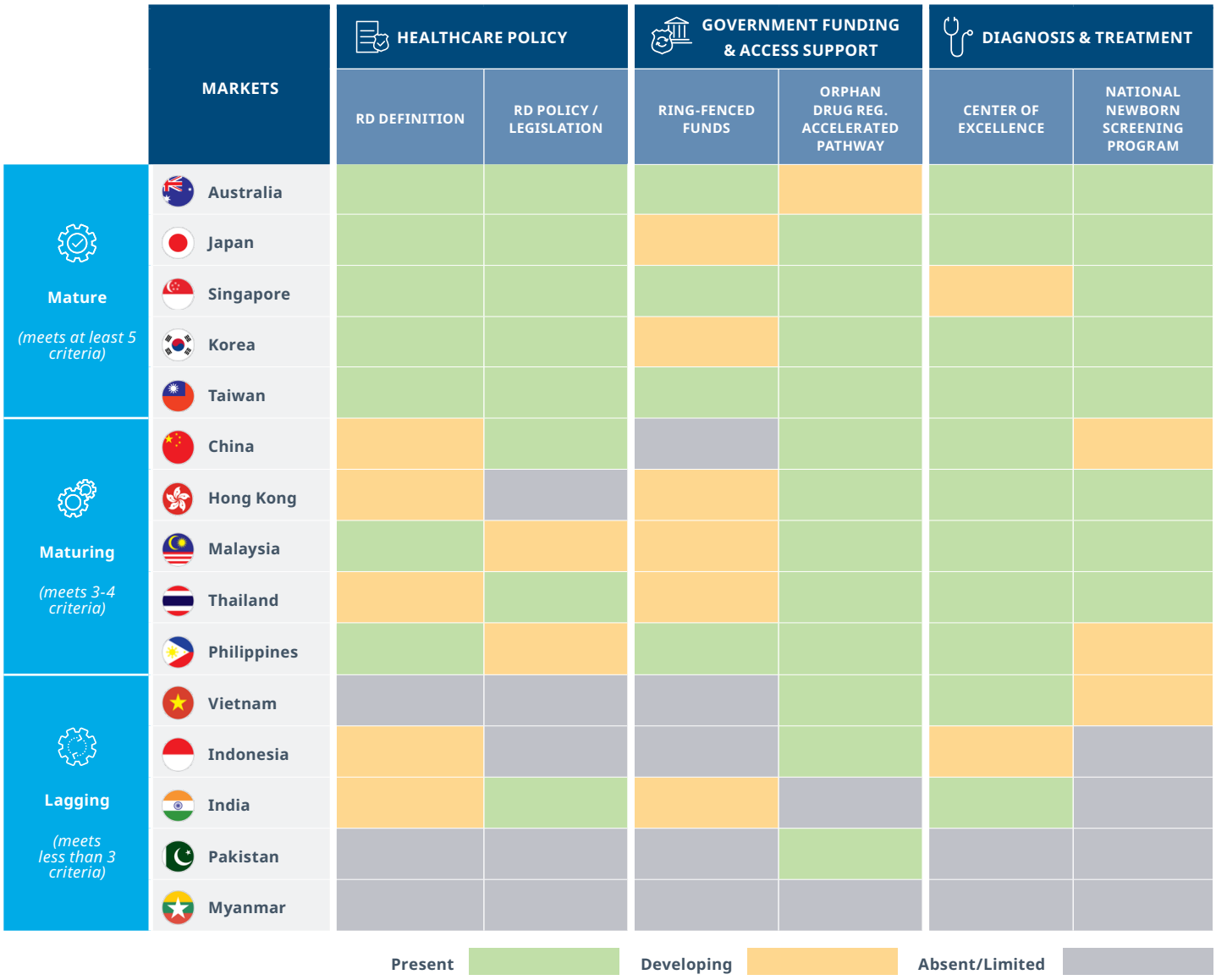
Based on our evaluation, we then divided APAC countries based on their level of maturity, i.e. mature markets, developing markets, and lagging markets, using the following logic:

Mature markets: Countries that meet 5 or more criteria (e.g. Japan, Taiwan)

Maturing markets: Countries that meet 3–4 criteria (e.g. Malaysia, Philippines)

Lagging markets: Countries that meet less than 3 criteria (e.g. India, Indonesia)

Figure 4: Maturity of APAC markets in terms of access to rare disease treatment



Source: IQVIA analysis, IQVIA expertise, Secondary Market Research [1, 2, 5, 7]

Among all the countries analysed, more developed markets such as Australia, Japan, Korea, Singapore, and Taiwan met almost all the criteria. Of these, Taiwan had one of the best models of governance for comprehensive rare disease management. Taiwan's Rare Disease and Orphan Act, enacted in 2000, included ~10-year exclusive marketing rights for orphan drugs and financial subsidies. Moreover, all Taiwanese patients affected by rare diseases are eligible for ~70% reimbursement on orphan drugs, with low-income patients eligible for 100% reimbursement. Lastly, patient advocacy groups in Taiwan provide strong support for patients with rare diseases.

Most countries in Southeast Asia (SEA) are still in the 'maturing' stage. Despite having made considerable progress towards achieving universal health coverage, the focus has largely been on preventative and primary health. Thus, funding for research and treatment of rare diseases is not prioritised and remains a challenge.

- **Thailand** represents a government-driven market with recent developments made to the rare disease landscape, with its Rare Disease Policy established in 2020 and the National Health Security Office agreeing to establish ring-fenced funding to cover ~24 rare diseases under its UCS.
- The **Philippines** enacted their Rare Disease Act in 2015 with strong support from patient advocacy groups, who played an important role in mapping out the national strategy. It is one of the first SEA countries to establish a national strategy dedicated to rare disease management.
- Countries such as **Malaysia**, on the other hand, are still developing their national policies on rare disease management. Malaysia is constructing a national framework rooted in coordinated activity, promoting timely diagnosis and treatment through multidisciplinary care of patients. Progress has been made since 2012, after orphan drugs were included as a part of the National Medicine Policy.

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Healthcare policy

Overall, there has been an increased focus on rare diseases in the APAC region, with policy developments reflecting emphasis on the rare disease agenda. A few prominent developments include:

- **Australia:** The government launched the country's first National Strategic Action Plan for Rare Diseases in February 2020.
- **Japan:** One of the first countries in the region to implement a dedicated policy since the 1970s; the country passed new legislation in 2014 and in 2015 founded the Agency for Medical Research and Development (AMED), which made rare and intractable diseases one of its priority areas.
- **South Korea:** In 2015, South Korea's Parliament passed the Rare Disease Management Act, requiring the Ministry of Health and Welfare to develop plans for the prevention, diagnosis, treatment, and research into rare diseases.
- **Taiwan:** The country's efforts towards rare diseases began in 2000 with the passing of the world's fifth Rare Disease and Orphan Drug Act. Since then, Taiwan has continuously improved care provision and, more recently, in 2017, published the Rare Diseases and Rare Genetic Disorders Care and Services Plan.
- **India:** The government of India formulated the National Policy for Treatment of Rare Diseases (NPTRD) in 2017, which initially faced implementation challenges and was eventually implemented in 2021.

Government funding and access support

Ring-fenced funds are dedicated, protected funding pools allocated for specific purposes and used by patients with rare diseases. These funds are not to be used or diverted for other causes and are exclusively reserved to support patients with rare diseases. Among APAC countries, the most mature markets have established ring-fenced funds, apart from countries such as Japan, where the government has a Drug Fund for Side-Effect Relief and Research Promotion that provides financial support for covering a part of expenses dedicated towards orphan drug research and development. Maturing markets such as Thailand and Malaysia are in the process of slowly expanding their funding for rare diseases, with limitations in therapy area coverage and disease scope.

Rare disease market definition

Neuromuscular disorders

- Generalized Myasthenia Gravis
- Spinal Muscular Atrophy (SMA)
- Duchenne muscular dystrophy (DMD)
- Huntington's Disease
- Achondraplasia
- hATTR Amyloidosis
- Neuronal ceroid lipofuscinosis type 2 (CLN2)

Blood / hematologic disorders

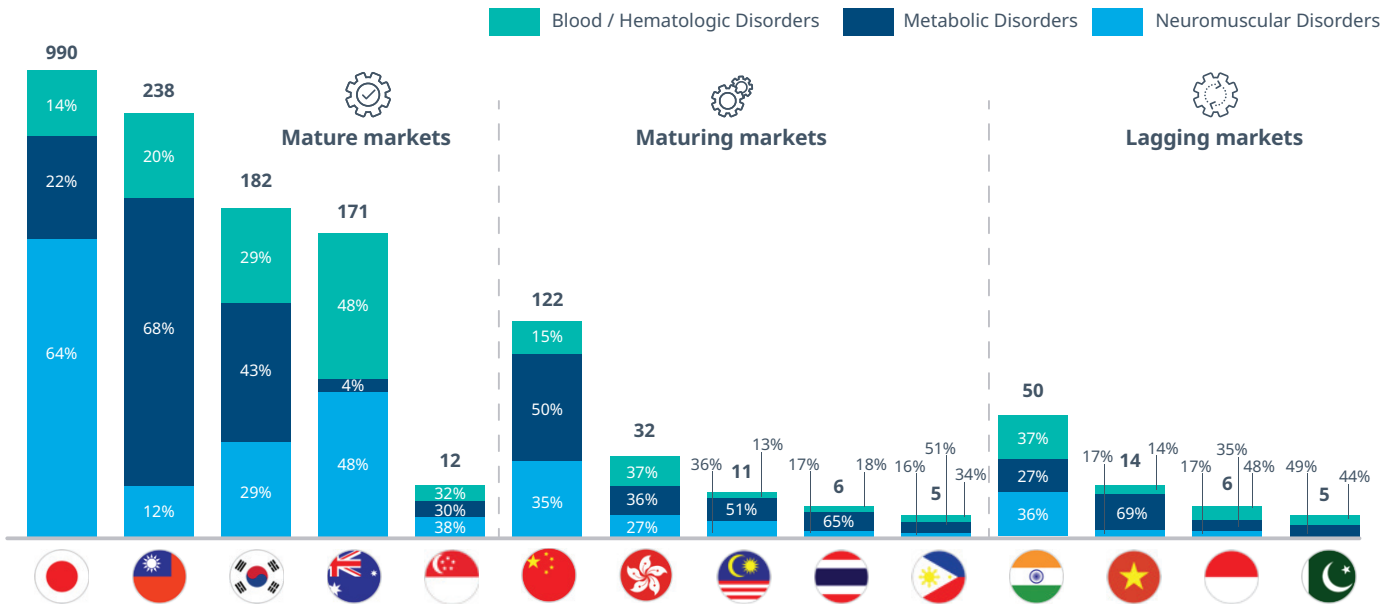
- Hemophilia
- Paroxysmal nocturnal hemoglobinuria (PNH)
- Hemoglobinopathies
- Factor Deficiencies
- Hypophosphatemic Rickets
- Hereditary Angioedema
- Idiopathic Cardiomyopathy

Metabolic disorders

- Gaucher disease
- Fabry disease
- Pompe disease
- Mucopolysarcharidosis (MPS)
- Hunter’s disease
- Nephropathic Cystinosis
- Phenylketonuria (PKU)
- Alagille Syndrome

Source: IQVIA expertise

Figure 3: Rare disease market size, segmented by disorders

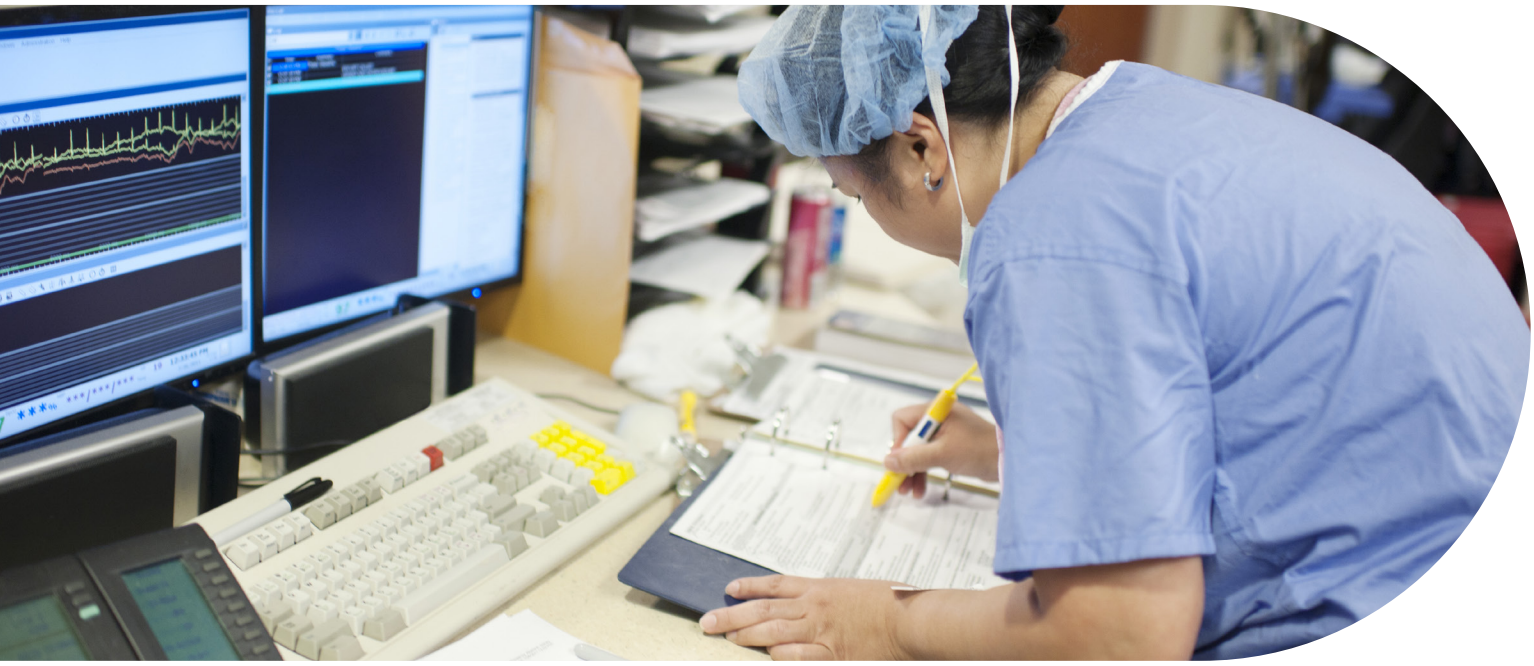


Source: IQVIA MIDAS data

Most countries in the APAC region have also established priority regulatory approval pathways for orphan drug accelerated registration meant to treat rare diseases, which are critical in bringing therapies to patients faster by considerably shortening the time to approval. The only outlier among the more developed markets is Australia, where benefits for orphan drug designation are limited. For example, only a waiver of registration fees for orphan drugs is offered. Recommendations to the Therapeutic Goods Administration (Australia's main regulatory authority for medications and medical devices) to provide orphan drugs with automatic access to the Priority Review Pathway were rejected by the Australian government in 2023. As of today, the Priority Review Pathway is granted only to medications for new prescriptions or indications used to treat, prevent, or diagnose life-threatening conditions and that have substantial evidence demonstrating major therapeutic benefit.

Initiatives to improve diagnosis and treatment

Almost all countries have established designated centres of excellence (or expertise) that house rare disease experts and serve as key centres for the diagnosis and treatment of rare diseases. These centres aim to specifically provide quality care to rare disease patients through cooperation and collaboration across specialised experts and clinical departments while also contributing to research. However, the number and scale of these centres differ greatly across the region, with countries such as Taiwan and Korea having a more widespread distribution across the country. Progress is being made in countries such as India, which is looking to expand its current network of 12 treatment centres and five diagnostic centres. Indonesia, too, opened its very first centre of excellence for rare diseases in Cipto Mangunkusumo Hospital in collaboration with the Human Genetic Research Center and the Indonesia Medical Education and Research Institute, Universitas Indonesia (IMERI UI).



Additionally, most countries, except for lagging markets such as Indonesia, have established national newborn screening programmes with varying degrees of funding, participation, and implementation. In China, newborn screening began in the 1980s, and screening techniques have evolved in the past few decades to include genetic testing. For countries in the SEA region, many tests need to be sent outside the country for analysis. Below are a few examples of newborn screening programmes in the SEA region:

- Thailand and Singapore screen close to ~100% of their newborns, while Vietnam and the Philippines screen only ~28%–30%. Newborn screening in the Philippines has been covered under national insurance since 2019.
- Newborn screening began in the 1980s in Malaysia, and the country has since expanded newborn screening efforts to include inborn errors of metabolism. However, these expanded tests are only available at two centres (Centre for Advanced Analytical Toxicology Services and Institute for Medical Research).
- In Singapore, more than 25 metabolic-related screening tests for newborns (including inborn errors of metabolism) are offered under the National Expanded New-born Screening Programme. Yet, more complex tests, such as those for lysosomal storage diseases, still need to be sent to more advanced countries (e.g. Taiwan) that have the required screening facilities in Asia.

APAC Rare Disease Market Sizing

In addition to understanding market maturity in the APAC region, knowledge about the relative rare disease market size of countries can help in prioritising the implementation of market access initiatives from a pharmaceutical industry perspective. Market sizing will also help indicate the level of penetration into the rare disease space by the pharmaceutical industry in each country and provide insights into therapy areas of focus.

In this section, we present an estimate of the rare disease market size in the APAC region and conduct a cross-country comparison of market size versus regulatory complexity to assess the potential for implementing market access initiatives and guiding market entry decisions to introduce new therapies in the region. By evaluating the regulatory complexity dimension, we can help indicate how well individual governments can support market access initiatives (e.g. the introduction of new therapies).

What is the size of the rare disease market in the APAC region?

We leverage IQVIA's proprietary market data and supplement them with other data sources (trade data, named patient programmes, etc.) to estimate the size of the rare disease market in the APAC region using the following market definition:

Rare diseases in the oncology space were excluded from our market definition, given that these may or may not be considered rare across all countries due to varying definitions. In addition, these are better tracked and more well-defined as compared to other paediatric and adult rare diseases.

Figure 3 illustrates the rare disease market size and indicates relative contributions from blood, neuromuscular, and metabolic disorders.

Japan leads the charts with a ~990 Mn United States dollar (USD) market, contributed mostly by the blood/haematologic disorder therapy area (~55%). Taiwan and South Korea are the next two most mature markets in order of market size, where treatments for rare metabolic disorders are the top contributors. This is also true for other maturing markets such as China, Malaysia, Thailand, and Vietnam, where therapies for rare metabolic disorders form the biggest chunk of sales, particularly those belonging to the lysosomal storage disorder group of diseases such as Gaucher disease, Fabry disease, and Pompe disease. Among the maturing markets, China is the clear leader, and India drives the lagging market segment with the maximum number of treatment options, compared with other countries. The latest audited data for Myanmar are not available due to the ongoing political unrest in the country; however, experts in the region indicate that the rare disease market can be valued at less than 500K USD, almost entirely driven by therapies for rare blood disorders.

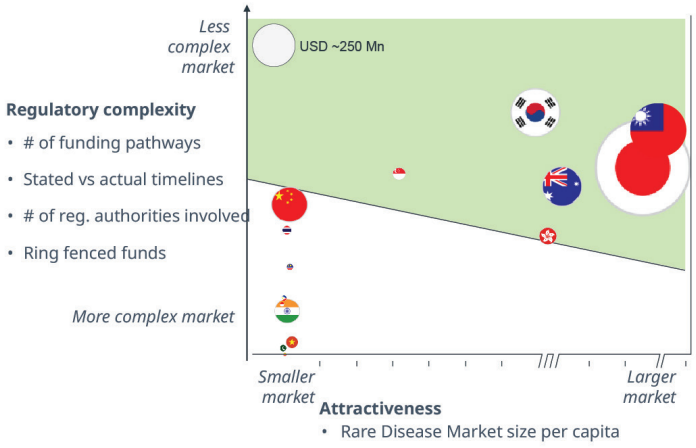
While the relative market sizes provide a good picture of the market potential, it is also essential to note the market size per capita to better understand how well the industry currently caters to the needs of the population. Thus, we next attempt to use the market size per capita metric, along with other metrics, to define regulatory complexity and identify markets that are better

equipped to support rare disease initiatives. *Figure 4* depicts a cross-country comparison, indicating that markets such as South Korea, Taiwan, and Japan are the most advanced markets, with rationales as follows:

- Ease of regulations through well-defined orphan drug pathways
- Multiple incentives defined for orphan drugs to enable quick market access (accelerated timelines, conditional approval, risk-sharing agreements)
- Well-connected network of support centres (~12) dedicated to rare disease diagnosis, treatment, and patient support
- Mature regulatory landscape with fast-track registration, trial waivers, and extensive comprehensive genetic disease prevention and control programmes
- Strong patient advocacy group influence on rare disease legislation and reimbursement
- One of the first countries to establish dedicated rare disease policies to clearly define a priority pathway for orphan drugs (preferential review) and also outline medical subsidies for rare disease patients

Figure 4; Prioritization matrix

Market Attractiveness (Prioritization Matrix)



Source: IQVIA MIDAS data, World Bank, Secondary Market Research [1, 2, 5, 7]

Conclusion

As home to the largest population of PLWRD (more than 250 million), it is heartening to see dedicated policies and legislation on rare diseases in place in almost all countries in the APAC region. However, the focus and extent differ across key aspects of rare disease management, ranging from research, awareness, diagnosis, and patient support to treatment access. Challenges remain in efforts to provide adequate care to PLWRD, such as the absence of a unified rare disease definition, a lack of epidemiological data, and poor healthcare practitioner awareness (which we explore in Part 2 of this white paper).

On the bright side, significant developments to improve access are underway across the APAC region, both among the higher-income and more technologically advanced countries/regions such as Japan, Korea, Taiwan, Hong Kong, China, and Australia, as well as developing markets such as India, Malaysia, the Philippines, Indonesia, and Vietnam. With governments taking initiatives to accelerate regulatory processes and play a greater role in funding treatment, the pharma industry can serve as a key stakeholder in bringing more treatment options to the region. The growing interest in improving access and reducing regulatory complexity in the rare disease landscape presents significant opportunities to fill unmet needs.

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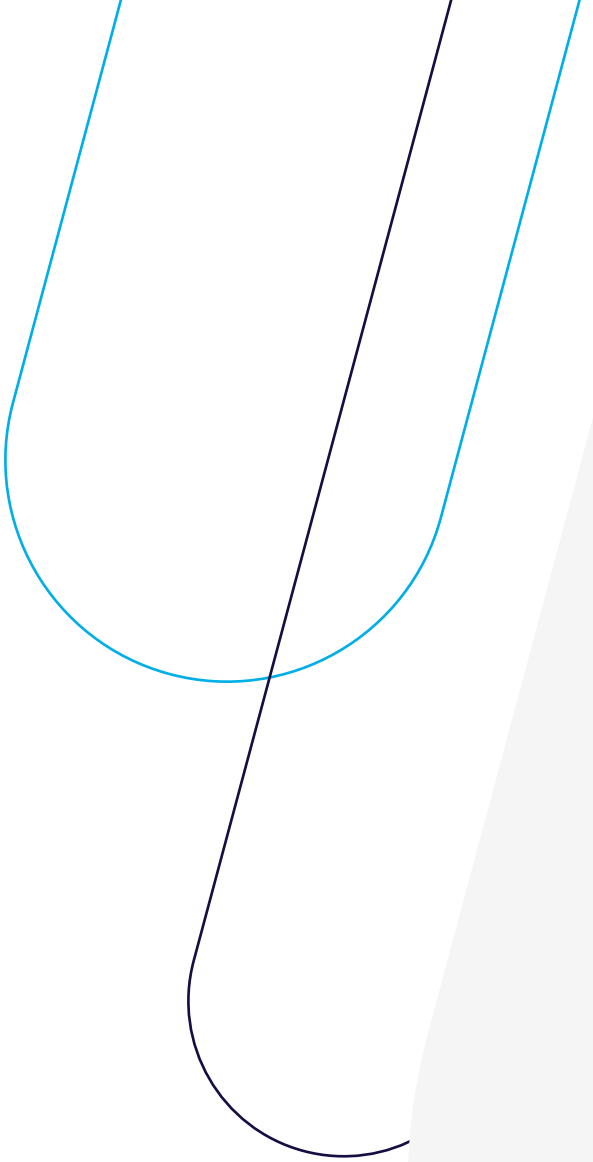

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