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The Socioeconomic and Quality of Life Impact of Living with Rare Disease Across Five Asian Countries

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Background

Existing economic evidence on rare diseases (RDs) in Asia is fragmented and drawn largely from **single-condition, single-country studies that capture medical costs only.[1]** No prior study has comparatively quantified the **cross-condition, multi-country socioeconomic impact** of RDs, spanning caregiver, productivity, mortality and quality-of-life effects, for this region.

Objective

To provide the **first cross-country assessment of the socioeconomic and quality-of-life impact of RDs** across diverse Asian health systems, quantifying impact at the **per-patient and national level** for **58 RDs across 5 Asian countries** (Malaysia, Singapore, South Korea, Thailand, Vietnam), and to compare the burden with that observed in a reference group, 70% of whom reported common chronic conditions such as diabetes.

Methods

Survey

Cross-sectional cost-of-illness analysis covering 58 RDs selected to span therapeutic area, age of onset and treatment availability (initial list from the literature, refined with clinical experts; illustrative, not exhaustive). Based on a structured survey of 523 PLWRD or caregivers responding on their behalf (of whom 59% were caregiver respondents) and 510 reference-group respondents, recruited via online panel in March 2026. The reference-group sample comprised people without an RD, 70% of whom reported common chronic conditions such as diabetes, providing a conservative comparator for estimating excess burden (not individually matched).

Literature review and stakeholder engagement

A targeted review of published impact studies, policy documents and epidemiological sources across all five countries served two purposes: to characterise the evidence gap and structural challenges facing PLWRD, and to supply the external model inputs, with country-specific estimates used where available and regional or international sources applied otherwise.

This was complemented by 22 semi-structured interviews (12 clinical, 10 policy experts) and engagement with **local patient advocacy groups**, Malaysia Rare Disorders Society, the Thai Rare Disease Foundation, KORD (South Korea), Rare Disorders Society (Singapore) and DEBRA (Singapore), and VORD (social enterprise, Vietnam), coordinated via Rare Diseases International.

Table 1: Survey sample size

Country	People living with rare diseases (PLWRD)	Reference
Malaysia	94	92
Singapore	128	108
South Korea	91	117
Thailand	92	103
Vietnam	118	90

Analysis and national extrapolation

Costs were estimated from a societal perspective, covering direct medical, direct non-medical, productivity and mortality-related costs. Survey-reported outpatient use covered 6 months and was annualised; inpatient use covered 12 months. Country-specific unit costs were applied to resource use, including caregiver-reported patient use. Productivity losses from absenteeism, presenteeism, reduced hours, early retirement and labour-force exit among PLWRD and caregivers were valued using the human-capital approach. Mortality losses were estimated separately using a human-capital model. Health-related quality of life (HRQoL) was measured using the EuroQol 5-Dimension 5-Level (EQ-5D-5L) instrument, and costs were adjusted using purchasing power parity (PPP). Per-person costs were pooled across five countries by therapeutic area and applied to country-specific prevalence. Adjusted scenarios are associational, not causal.

Results

The impact of RDs on society is significant

Five Asian countries incur excess annual costs of PPP US$63.8 billion across 924,000 people living with any of 58 RDs in scope; each person carries PPP US$69,000 in excess cost per year. This shows RD have a cost 3-4 higher than the reference group. On average across the five countries, RDs impose around 4 times the excess cost of diabetes (PPP US$18,400).

Table 2: Cost composition by country (% of excess total cost)

Cost category	Malaysia	Singapore	South Korea	Thailand	Vietnam	All
Direct medical	57	55	60	52	58	**56**
Direct non-medical	22	21	22	19	21	**21**
Indirect (productivity)	12	13	13	10	12	**11**
Mortality-related	8	11	5	19	8	**12**

Direct medical care accounted for the largest individual share of excess cost. However, direct non-medical, productivity and mortality-related costs together represented 44%, demonstrating that a substantial proportion of the socioeconomic impact falls outside healthcare budgets. Travel costs may partly reflect the centralisation of specialist RD services identified through stakeholder engagement (Table 2).

Table 3: Excess impact by therapeutic area (total and per person)

Therapeutic area	Total burden (PPP US$, billions)	Cost per person per year (PPP US$)
Haematologic	US$19.8	US$86,000
Neurologic	US$13.4	US$71,000
Immunologic	US$11.4	US$56,000
Congenital malformations	US$7.0	US$52,000
Metabolic	US$6.2	US$116,000
Endocrinologic	US$5.3	US$55,000
Cardiovascular	US$0.7	US$44,000

Table 3 demonstrates the high level of heterogeneity between different therapeutic areas. This ranges from US$116,000 for metabolic diseases to US$44,000 for cardiovascular diseases. Although the sample size is too small to report individual diseases there is clearly even more heterogeneity at the disease level.

The diagnostic odyssey

34% **of PLWRD were misdiagnosed** at least once in their diagnostic journeys, annual costs were 2–3 times higher among those reporting misdiagnosis

1.4x **more likely to be misdiagnosed** in rural areas than in urban areas

up to 44 GPs and 24 specialists **are seen by PLWRD** before receiving the correct diagnosis (averaging to 2 for both categories)

50–83% **past-year genetic testing rate among PLWRD with genetic conditions** (ranging from Vietnam to Singapore), showing regional inequity in diagnostic access

2 years **mean time to diagnosis, ranging from 1 month to 20 years;** time to diagnosis is 3 times longer when treatment is not available at time RD onset

Financial impact on PLWRD households

47% **of PLWRDs spend above the World Health Organization catastrophic-spending** threshold (>25% of income spent on health-related items)[2]

57% vs 37% **share of direct costs paid out-of-pocket by RD versus reference households,** indicates greater exposure to uncovered costs

24% **lower HRQoL for PLWRD than the reference group** (lowest for neurologic conditions)

67% **of PLWRD require two caregivers** compared to 20% of reference group respondents

Access to treatment and care

89% **face at least one barrier to reaching care,** including limited specialist appointments, slow authorisation of services, or no nearby RD specialist

45% **longer travel time** to care for rural than urban patients

4x **higher emergency care use among PLWRD vs reference group;** suggesting that gaps in specialist care may contribute to reliance on acute services

>5 months **the average wait to start treatment after diagnosis, ranging up to 20 years** potentially delaying effective management

74% **report treatment-access challenges** driven by affordability and authorisation rather than clinical suitability

Wider impact on society & economy

59 days **of productivity lost** by PLWRD on average, with losses reaching a full working year for some leaving employment; 75% reported family-related stress, while caregivers lost around 66 days of productivity

3x **more likely to miss education,** interrupting development and future earning potential

US$7.7 billion **in productive potential lost** annually through premature mortality

77% **of RD caregivers miss work to attend appointments;** they were **2x as likely** as non-RD caregivers to report their **health has suffered due to caregiving responsibilities**

Modelled scenarios: potential cost offsets from intervention

- Scenarios focused on modifiable features of the diagnostic and care pathway that were associated with higher annual costs across the five countries and could be robustly parameterised using the available data. Clinically important mechanisms that could not be modelled reliably were not quantified. The offset represents the estimated difference in annual cost if the specified pathway feature were changed, with other modelled characteristics held constant.
- The largest offsets were associated with halving pre-diagnosis provider contacts and halving emergency visits, followed by eliminating misdiagnosis. Improvements in HRQoL and reduced disease-related travel were also associated with lower annual costs.
- The concentration of the largest offsets around provider contacts, emergency care and misdiagnosis suggests that earlier and more efficient diagnosis, alongside better coordinated care, may offer important opportunities to reduce costs.
- The HRQoL and travel scenarios indicate that potential value may also extend beyond direct healthcare expenditure to outcomes affecting PLWRD and their families.

Table 4: Illustrative annual cost offsets associated with modelled counterfactual scenarios

Modelled mechanism (counterfactual)	Cost offset (PPP US$/person/year)
Halving pre-diagnosis provider contacts (including GPs and specialists)	**21,600**
Reducing emergency department visits by half	**17,800**
Eliminating misdiagnosis	**9,800**
Each +0.1 gain in HRQoL (EQ-5D utility)	**9,200**
Halving disease-related travel costs	**6,700**

Conclusion & Limitations

Key conclusions

1. Across the 58 RDs studied, an estimated 924,000 PLWRD were associated with PPP US$63.8 billion in excess annual socioeconomic costs across five Asian countries.
2. Average excess annual cost per PLWRD was approximately 3 times that observed for diabetes, reflecting financial, productivity and HRQoL effects beyond direct medical expenditure.
3. The largest illustrative cost offsets were associated with fewer pre-diagnosis provider contacts, reduced emergency care and avoidance of misdiagnosis, although these scenario findings are associational rather than causal.

Avoidable costs and inefficiencies arise across the RD patient journey. Earlier diagnosis, coordinated care and timely treatment, supported by stronger financial protection, caregiver support and accountable data systems, should therefore be priorities for further investment and evaluation.

Ethics, funding, and acknowledgements

- **Ethics:** All survey and interview participants provided informed consent before taking part. Data were collected and analysed in de-identified and aggregated form, in accordance with applicable data protection requirements.
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Limitations

- **Cross-sectional design:** associations are not causal; scenario offsets are illustrative.
- **Online-panel recruitment** may under-represent the most severely affected or marginalised PLWRD, likely making estimates conservative. Self-reported resource use is subject to recall bias.
- **Prevalence uncertainty**: in the absence of national registries, national totals are indicative of the 58 conditions studied, not the full RD population.
- **Reference group** includes common chronic conditions rather than a matched general population.

Abbreviations: EQ-5D-5L, EuroQol 5-Dimension 5-Level; GP, general practitioner; HRQoL, health-related quality of life; ISPOR, International Society for Pharmacoeconomics and Outcomes Research; KORD, Korean Organization for Rare Diseases; PLWRD, people living with rare diseases; PPP, purchasing power parity; RD, rare disease; US$, United States dollars; VORD, Vietnamese Organisation for Rare Diseases; WHO, World Health Organisation.

[1] Shafie, A. A., Chaiyakunapruk, N., Supian, A., Lim, J., Zafra, M., & Hassali, M. A. (2016). State of rare disease management in Southeast Asia. Orphanet journal of rare diseases, 11(1), 107. https://doi.org/10.1186/s13023-016-0460-9; [2] World Health Organization. (n.d.). Financial hardship: Population with household expenditures on health greater than 25% of total household expenditure or income (SDG indicator 3.8.2). https://www.who.int/data/gho/indicator-metadata-registry/imr-details/4845

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