The socioeconomic impact of rare disease
An analysis of the evidence in middle-income countries

Over 300 million people are living with one of the **6,000-8,000 rare diseases** identified to date

This research describes the socioeconomic impact of certain rare diseases[1] on persons living with a rare disease, families and society, with a focus on middle-income countries (MICs)[2]

The approach followed five steps:

Develop research framework

Select rare diseases[1] and countries[2]

Consultation sessions with IFPMA and Rare Diseases International (RDI) representatives

Conduct literature review, collect data

Evaluate estimates, note limitations, draw conclusions

Finding 1

The estimated socioeconomic impact of rare diseases is significant regardless of country income level

Diseases like hemophilia and multiple myeloma have a similar socioeconomic impact per diagnosed and treated patient across MICs and comparator high-income countries (HICs)

Finding 2

Prevalence of rare diseases is underestimated and therefore less visible in MICs

30 million rare disease cases were estimated to be unreported across the diseases and MICs studied

Finding 3

Existing information on the socioeconomic impact of rare diseases is limited in MICs

Direct medical costs[3] and indirect costs[4] can be quantified but there are issues with data quality and extrapolation is required

Costs falling directly on the patient[5] only assessed qualitatively

Finding 4

Composition of direct and indirect costs varies by diseases and countries and is affected by:

Limited availability of and access to specialists

Differences in existence of clinical guidelines

Variation in access to optimal treatments

Finding 5

Impact on patient and caregiver experience remains critical, even if challenging to quantify

Outdated treatment options and delayed diagnosis for patients leads to:

- Higher mortality rates
- Greater disease progression and severity
- Reduced quality of life

Lack of policy support and financial compensation for caregivers leads to:

- Strain and mental health issues
- Social isolation
- Poor work-life balance

1. Gaucher disease, mucopolysaccharidosis II, hemophilia, idiopathic pulmonary fibrosis, multiple myeloma, myasthenia gravis 2. Brazil, China, Chile, Colombia, Egypt, Ghana, Kenya, Malaysia, South Africa, Thailand, and high-income comparator countries Australia, Taiwan. 3. Direct medical costs quantified included inpatient stay, outpatient care, and prescription medication. 4. Indirect costs quantified included absenteeism/presenteeism and early retirement for persons living with a rare disease and their caregivers. 5. Costs placed directly on the patient included mortality outcomes, quality of life impact, and transportation costs.

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Following the 2021 UN Resolution on rare diseases and the Member States' call for a WHA resolution and a global action plan on rare diseases, there is an opportunity to focus global policymakers on the specific needs of persons living with rare diseases

Policy recommendations

Prioritization of rare disease intervention is vital to address the significant impact identified, with specific actions to consider national contexts

Establish programs to improve screening and early diagnosis, as they have important clinical benefits and can reduce the socioeconomic impact of rare disease on persons living with a rare disease, caregivers, and societies

Improve the collection of data to ensure the complete socioeconomic impact of rare disease is understood and that policies can be designed appropriately

Invest in the training of specialists, improve healthcare provider awareness, and **establish national reference centers** to expand access to dedicated care

Ensure access to effective treatments for all patients to reduce the impact on persons living with a rare disease, their caregivers, and on other parts of the healthcare system

This study shows that the scale of the socioeconomic impact of rare diseases in MICs per patient is significant. However, significant data gaps mean that we cannot develop aggregate estimates of the socioeconomic impacts as in other regions. More robust and granular data is needed to understand the scale and composition of the socioeconomic impact of rare disease in different healthcare settings

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IFPMA, in collaboration with Rare Diseases International (RDI), engaged Charles River Associates (CRA) to investigate the broad socioeconomic impact of rare disease, drawing upon the evidence for a diverse set of conditions, with a focus on lower-middle and upper-middle-income countries. The data collection and analyses were conducted by the CRA team with regular feedback from IFPMA members and RDI representatives. Additional guidance was provided by Professor Steven Simoens (Professor of Health Economics at KU Leuven, Belgium).

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