Exploring the Development of a Framework for Prioritizing Rare Inherited Disease Research in Low- and Middle-Income Countries

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Current Positions:
(1) Deputy Director (Commercialization and Support Services), Institute for Research, Development and Innovations (IRDI), IMU University, Malaysia;
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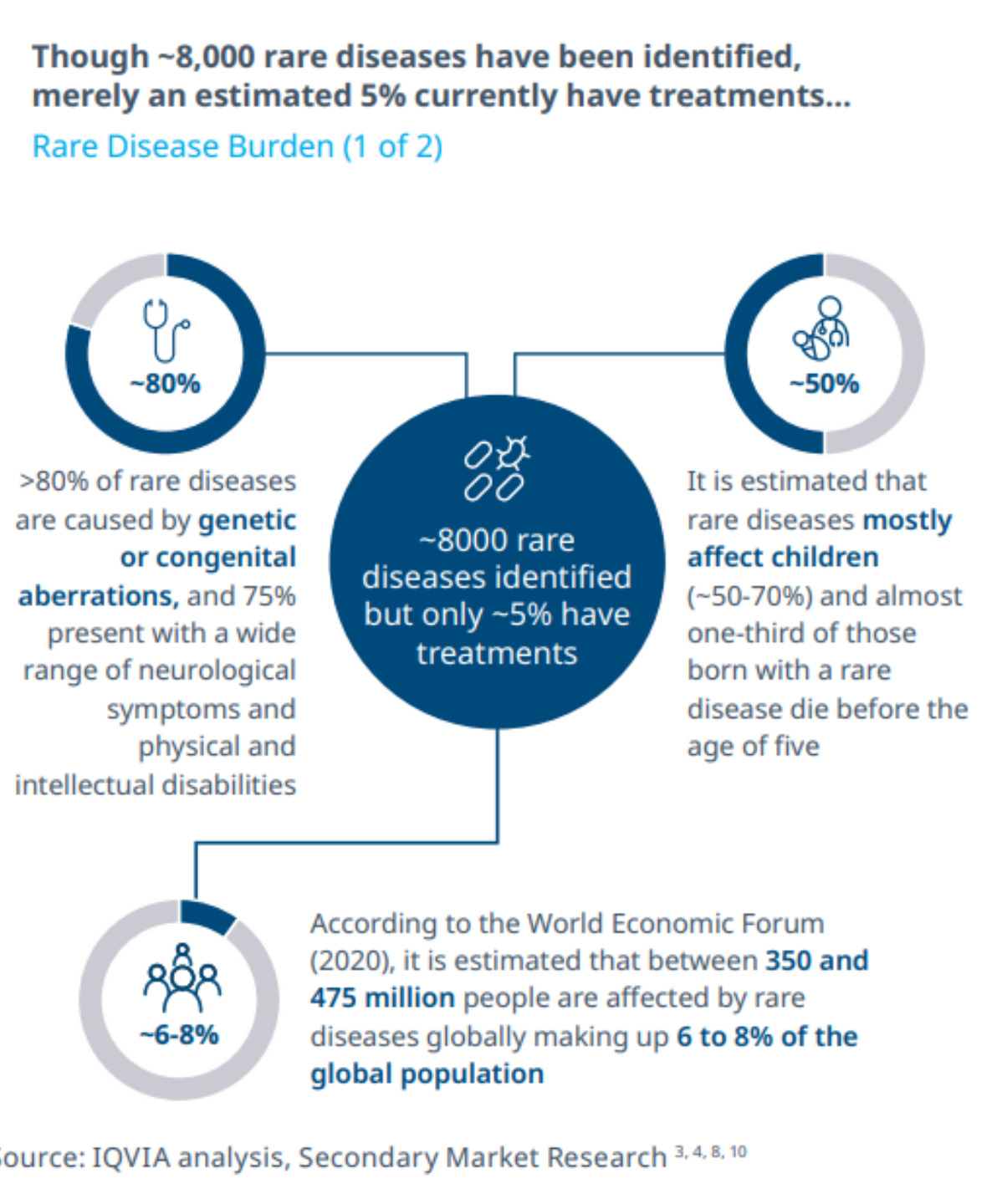
Dr. Teguh brings over fifteen years of expertise in human genetics, bioethics, and healthcare research. His work centers on developing ethical frameworks for health research prioritization in low- and middle-income countries (LMICs), with a focus on integrating rare diseases into national health research strategies. Dr. Teguh has authored numerous peer-reviewed publications across the fields of human genetics, bioethics, and evidence-based healthcare, with a recent emphasis on ethical inclusion for rare disease research in LMICs. He has secured substantial research funding from institutions such as the Global Forum on Bioethics in Research (WHO-NIH) and the Malaysian Ministry of Higher Education, supporting projects on rare disease prioritization, AI-powered diagnostics, and genetic epidemiology in Southeast Asia. His research initiatives include culturally attuned frameworks assessing disease severity and community vulnerability to better serve marginalized populations.
In recognition of his contributions, Dr. Teguh has held visiting research positions in the United States, Japan, Indonesia, and has an upcoming role in the United Kingdom. These roles reflect his international engagement and dedication to advancing ethical, equitable healthcare across resource-limited settings in Southeast Asia.

Confronting the Overlooked Crisis of Rare Diseases

- **Definition:** Rare diseases affect <1 in 2,000 individuals
- **Prevalence:** 6-8% of global population
- **Thalassemia:** Lifelong treatment, significant financial burden
  - SEA Prevalence: ~17% - ~50%
- **Spinal muscular atrophy:** Long diagnosis journey, lack of effective treatments



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Strategic frameworks are crucial for effective resource allocation

Neglect of rare diseases in global health research and LMICs.

Importance of prioritization in resource-constrained settings.

Payer	Gaps identified locally
Government	Insufficient **RD fund allocation** Lack of **RD strategy** (short or long-term) Absence of **national governing body** or **committee** for rare disease **Absence of legislation** for adoption of funding programs Poor **RD awareness**
Private insurance	Absence of **RD coverage policy** or requires **high co-payments** No **coverage expansion incentive** for RD
Out-of-pocket	Low ability to fund high-cost innovative drugs, often leading to financial catastrophe

Country	Rare disease maturity of the public sector			
	Ring-fenced funds	RD patient registry	Orphan drug registration pathway	Accelerated registration pathway
Romania	✓ USD 100 Mn	✓		
Thailand	✓ <USD 1 Mn		✓	✓
Argentina		✓	✓	✓
Colombia				
Peru				
South Africa				
Lebanon				
Malaysia	✓ USD 4 Mn		✓	✓
Morocco				
Ghana			✓	✓

Roche-IQVIA 2021

✓ Present ✓ Partially present Absent

Research Goals and Objectives

Develop a framework to prioritize rare disease research in LMICs.

Address resource constraints, align with local needs, and reduce disparities.

Promote inclusivity, equity, and collaboration.

Challenges: Limited resources, alignment with local needs, global disparities.

Expected outcomes: Inclusivity, equity, and international collaboration.

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Framework Overview *(early draft)*

Five components:

1. **Identify candidate priorities.**
2. **Global prioritization criteria.**
3. **Local prioritization criteria.**
4. **Stakeholder engagement.**
5. **International collaboration.**

Ensure evidence-based, inclusive, adaptable framework.

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Identification of Candidate Priorities

Methods: Epidemiological data, stakeholder consultations, review of unmet needs.

- **Case Example**: Strong stakeholder contribution from the Philippines Society for Orphan Disorders (PSOD):
  - Drafted National Rare Disease Strategy.
  - Advocated for Rare Disease Act (2015).
- **Unmet needs in LMICs:** Awareness, diagnostics, funding, policy gaps.

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Criteria for Global Prioritization

Severity-weighted disease burden using DALYs.

Forecasting potential disease burdens.

Case Study: Netherlands

Dutch Cystic Fibrosis Registry (Nederlandse Cystic Fibrosis Stichting) Available at: https://ncfs.nl.

Dutch Dystrophinopathy Database (Duchenne Data Foundation. *Facilitation of Trial Readiness and Effective Use of Patient Data*. Available at: https://www.duchennedatafoundation.org.

DALYs and Rare Disease Prioritization
Cleemput I, Neyt M, Thiry N, Van de Sande S. The use of DALYs in health economic evaluations: A quantitative study. *Archives of Public Health*. 2020;78:68. Available at: https://archpublichealth.biomedcentral.com.

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Criteria for Local Prioritization

Focus on local disease burden and alignment with national priorities.

Case Study: Ghana's National Sickle Cell Disease Strategy (2024–2028) (ghanatoday.gov.gh):

- Comprehensive prevention, diagnosis, and treatment under NHIS.
- Pneumococcal vaccination for SCD patients.

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Stakeholders Engagement

Roles: Patients, providers, researchers, policymakers contribute to prioritization.

Case Study: Philippines Society for Orphan Disorders (PSOD)

- o Advocacy, Rare Disease Act, collaboration with NIH-UP Manila on a national registry.

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International Collaboration

Importance of resource sharing with high-income countries.

Strategies for LMIC engagement: Public-private partnerships, leveraging international aid.

Example: Cross-border genetic screening between various LMICs - HICs.

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What we plan to do next

Steps:

1. **Launch pilot programs in LMICs.**
2. **Build capacity through training.**
3. **Raise awareness among policymakers and communities.**

Focus on sustainable ecosystems for rare disease research.

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Challenges and Opportunities

Challenges:

- Political and financial barriers

- Ethical dilemma in prioritization

- Ensuring sustainability.

Opportunities:

- Data-driven approaches like DALYs for adaptability and resource allocation.

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Conclusion

Importance of aligning research with health equity goals.

Call to action: Collaboration, inclusivity, and strategic prioritization.

Vision: A framework that ensures equitable access to innovations for rare disease patients.

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World Health Organization

NIH National Institutes of Health
Turning Discovery Into Health

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INYUVESI YAKWAZULU-NATALI

Global Forum on Bioethics in Research

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Thank you for the funding!

Thank you.

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