

## Understanding the Co-Benefits of Rare Disease Policy, Planning, and Programs

*“When we solve problems for rare diseases, we strengthen health systems as a whole”*

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### Introduction

The concept of “co-benefits” originated in the climate science community to describe the added advantages gained from actions taken to control climate change, above and beyond the direct benefits of a more stable climate. For example, a city’s investment in bicycle lanes reduces greenhouse gas emissions, but also encourages healthier lifestyles and reduces incidences of non-communicable diseases linked to physical inactivity. Co-benefits can also be thought of as ripple effects or dividends. Similarly, developing and investing in national rare disease (RD) policies, plans, and programs generate many co-benefits. By addressing the unique challenges posed by RDs, national policies, plans, and programs and their impacts help not only improve the lives of people living with a RD, but also strengthen healthcare systems, enhance research and innovation, and improve outcomes for more prevalent conditions like cancer, cardiovascular disease, and infectious diseases.

Understanding and effectively communicating these co-benefits is crucial for building political will and addressing concerns related to resource allocation. Policymakers often face difficult decisions when prioritizing limited resources, and RD initiatives may be perceived as more costly, too narrow, or less urgent. However, by highlighting the broader benefits of RD policies—such as their potential to drive innovation or improve healthcare infrastructure—advocates can demonstrate that these investments serve multiple priorities. Additionally, RD policies align with broader public health goals, such as improving national health outcomes and advancing equity in healthcare access, making them a strategic component of sustainable development and overall healthcare improvement. This approach helps counter objections related to resource scarcity and positions RD programs as a strategic investment with widespread impact for everyone.

### Key Co-Benefits of Rare Disease Policy, Planning, and Programs

1. Generate savings in the healthcare system and foster economic growth
2. Enhance medical research, innovation, and clinical care for more prevalent health conditions
3. Improve diagnostics and screening for cancer and infectious diseases
4. Encourage interdisciplinary collaboration in clinical medicine, academic research, and policymaking
5. Enhance healthcare infrastructure
6. Foster health equity

### 1. Generate Savings in the Healthcare System and Foster Economic Growth

Investing in RD programs can lead to substantial healthcare savings by improving early diagnosis, preventing complications, and optimizing resource use, benefiting broader patient populations, and reducing system-wide costs, both direct and indirect.

- **Prevention of Disease Progression:** Early diagnosis and treatment of RDs prevent severe complications and reduce the need for expensive interventions. For example, treating metabolic disorders like PKU or lysosomal storage disorders early can avert organ damage and long-term disabilities, cutting down on costly surgeries, transplants, or intensive chronic care including in-home services and caregiver costs.
- **Reduced Hospital Admissions:** Effective management of RDs can minimize hospitalizations and emergency care, reducing overall healthcare utilization. Conditions like genetic immune deficiencies or bleeding disorders, when properly managed, avoid the frequent hospital visits that untreated cases might require, leading to more efficient use of resources.

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- **Reduced Social Care Costs:** Effective RD programs can lower the burden on social care systems by reducing disability rates and the need for long-term caregiving. Early diagnosis and treatment sometimes enable patients to maintain greater independence, minimizing reliance on disability benefits, in-home care services, and caregiver assistance.
- **Increased Workforce Productivity:** Successful treatment of RDs allows patients to remain productive members of society, reducing economic dependence. For instance, advancements in treating cystic fibrosis or spinal muscular atrophy extend patients' lifespans and quality of life, allowing them to participate in the workforce longer and reducing the caregiving burden on families.
- **Economic Growth through Innovation:** RD policies stimulate research and development, driving innovation in treatments and diagnostics that have applications in other areas of healthcare. Investments in RD diagnostics and therapies often lead to job creation and growth in the health and life science sectors, benefiting the wider economy.
- **Efficient Use of Healthcare Resources:** By optimizing early diagnostics and treatment pathways, RD programs reduce long-term healthcare costs. Technologies like patient registries and biobanks help accelerate research and development while reducing duplication of efforts, allowing resources to be used more effectively across the healthcare system.

### 2. Enhance Medical Research, Innovation, and Clinical Care for More Prevalent Health Conditions

Investing in RD research catalyzes and accelerates medical advancements and innovations that can be applied to more prevalent health conditions, including the discovery of novel therapies and scientific breakthroughs.

- **Cancer:** The study of RDs like Li-Fraumeni syndrome and retinoblastoma has advanced our understanding of cancer genetics, biology, and susceptibility, driving the development of targeted therapies and preventive measures for high-risk populations (e.g., BRCA1 and BRCA2). These advances now benefit more common forms of cancer.
- **Infectious Diseases:** Research on RDs like primary immunodeficiencies (e.g., severe combined immunodeficiency, chronic granulomatous disease) and rare genetic variants associated with resistance to HIV infection has provided insights into immune system functions and genetic factors that influence susceptibility to infections. This knowledge has benefited the management of immunocompromised patients, including those with infectious diseases like HIV.
- **Cardiovascular Diseases:** Investigations into familial hypercholesterolemia, a RD leading to high cholesterol, have led to the development of statins and more recently PCSK9 inhibitors, which are now staples in managing high cholesterol and reducing heart disease risk in broader populations.
- **Diabetes:** Research on rare forms of monogenic diabetes, such as maturity-onset diabetes of the young, has helped identify key genetic factors involved in diabetes development, informing both diagnostics and therapeutic interventions for type 1 and type 2 diabetes.

### 3. Improve Diagnostics and Screening for Cancer and Infectious Diseases

Developing RD policies, plans, and programs often requires improvement of and investment in diagnostic and screening technologies and infrastructure that benefit the broader medical field. RD diagnostics have can be applied to a broader range of health conditions and have spurred advancements in early detection and personalized medicine.

- **Genetic Testing:** Next-generation, whole-genome, and whole-exome sequencing, originally developed for identifying mutations in RDs, are now widely used in precision oncology to tailor treatments based on

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patients' unique genetic profiles. These tools are also applied to infectious diseases for identifying pathogens, understanding drug resistance, and tracing transmission patterns.

- **Laboratory Techniques:** Initially developed to study rare immunodeficiencies, flow cytometry is now a critical tool in diagnosing leukemia and lymphoma, and in monitoring immune responses in patients with HIV or other immune-related conditions.
- **Imaging Technology:** RDs have driven advances in imaging technologies, such as MRI, CT, and PET scans. These tools are now indispensable in diagnosing and tracking the progression of conditions like cancer and neurodegenerative diseases.
- **Data Sharing & Integration:** RD programs often foster the creation of biobanks and development of patient registries and data-sharing frameworks. These systems have proven valuable in improving epidemiological surveillance and research for diseases such as cancer and infectious diseases.
- **Genetic Counseling:** RD policies, plans, and programs often seek to expand and improve genetic counseling services, which help patients and families understand the genetic basis of their conditions. These services are increasingly being applied in cancer genetics and cardiovascular care to support informed decision-making.

#### 4. Encourage Interdisciplinary Collaboration in Clinical Medicine, Academic Research, and Policymaking

The inherent complexity of RDs demands collaboration across various medical specialties, research disciplines, sectors, and institutions. This interdisciplinary approach can lead to innovative solutions for other diseases.

- **Multidisciplinary Clinical Teams:** RD programs often involve healthcare professionals from multiple specialties, including geneticists, radiologists, clinical researchers, pharmacists, nurses, social workers, and patient advocates. These teams work together to provide comprehensive care for RDs, which can serve as a model and pilot for treating other complex diseases.
- **Collaborative Research:** RD research often requires partnerships between academia and industry, and across multiple academic departments. Such collaborations have broad implications for conditions such as oncology, neurology, and immunology, accelerating the development of new diagnostic tools and therapies, as well as the discovery of new genes and pathways involved in disease development.
- **Patient Participation in Research:** In RD research, patients and advocacy groups often play a significant role in shaping research agenda, offering unique insights into disease management and sometimes helping fund research. This approach can enhance and provide models for patient-centered research and engagement across the entire healthcare system.
- **Innovative Clinical Trial Design:** RD research often requires novel approaches to clinical trials due to small patient populations. Adaptive trial designs, basket trials, and umbrella trials—methods pioneered in RD—allow more flexible, efficient testing of therapies. These designs are now increasingly applied in broader fields, such as oncology and infectious diseases, accelerating drug development and approval processes.
- **Interagency Collaboration on Policymaking:** Developing RD policies and plans often requires collaboration across multiple government bodies, including health ministries, finance, education, and social services. This interagency coordination fosters better communication, streamlined policy implementation, and more holistic approaches to healthcare. As agencies work together on RD issues, they build frameworks that enhance policy development and program execution for other health challenges, such as national responses to pandemics or chronic disease management.

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- **Multistakeholder Participation in Policymaking:** RD policies often involve collaboration among diverse stakeholders, including patients, advocacy groups, healthcare providers, industry representatives, and policymakers. This inclusive approach ensures that policies are patient-centered while also leveraging industry expertise to drive innovation. This collaborative model can serve as a blueprint for addressing broader health challenges, fostering more equitable and effective health policy development.

### 5. Enhance Healthcare Infrastructure

Investments in RD programs often lead to substantial improvements in healthcare infrastructure, which can benefit the entire healthcare system. These investments drive the development of specialized facilities, tools, and systems that improve care delivery not only for RD patients but also for broader patient populations facing complex medical conditions.

- **Centers of Excellence:** RD programs often establish specialized centers that provide innovative diagnostics, treatment, and care management. These centers not only serve RD patients but also offer a model for addressing complex medical conditions, such as cancer or neurological disorders. The multidisciplinary approach and focus on advanced research and care in these centers can be replicated across healthcare systems to improve outcomes for a broader patient population.
- **Support for Caregivers:** Caring for patients with RD often places significant physical, emotional, and financial demands on caregivers. RD programs often offer support in the form of education, respite care, and mental health resources. These caregiver support structures can be expanded to benefit those caring for individuals with other chronic or complex conditions, ensuring they have the tools and assistance needed to provide effective, sustainable care.
- **Training and Education:** RD initiatives invest heavily in educating healthcare professionals on the complexities of diagnosing and treating rare conditions. This training enhances the overall capacity of the healthcare workforce, improving their ability to manage complex and emerging medical issues. Increased awareness and knowledge from these programs can also enhance care for patients with other challenging conditions, fostering a healthcare system that is more responsive to rare and common diseases alike.

### 6. Foster Health Equity

RD policy, planning, and programs are uniquely positioned to promote health equity and can drive broader reforms aimed at achieving more inclusive healthcare systems. By addressing the needs of RD patients, governments can deepen and demonstrate their commitment to health equity and ensuring that all individuals, regardless of their condition, have access to appropriate care and support.

- **Addressing Disparities in Access:** RD policies bring attention to gaps in healthcare access, highlighting the need to ensure that all populations, regardless of socio-economic status, have access to quality care. These policies set a precedent for addressing similar disparities in the treatment of common chronic diseases.
- **Affordable Access to Treatments:** RD policies emphasize making treatments affordable and accessible. This commitment to equitable treatment can inform policies that improve affordability and access to innovative therapies for more prevalent conditions.
- **Community-Based Care:** By bringing healthcare resources to underserved communities, RD programs offer a blueprint for expanding community-based care across a wide range of health conditions. This approach ensures that patients can receive care in settings that are accessible and responsive to their cultural and social contexts.