IFPMA

Leaving no-one behind: a set of policy principles to meet the global challenge of RARE DISEASES

INTRODUCTION

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Rare diseases pose unique challenges to patients, their families, society, healthcare professionals and healthcare systems. Globally, some 7,000 rare diseases have been identified. Positively, a number of significant events in the global policy agenda have helped to give more momentum to rare diseases, such as the UN 2030 Agenda and Sustainable Development Goals (SDGs) and the drive towards universal health coverage, which are centred around leaving no-one behind, as well as the recent establishment of the NGO Committee for Rare Diseases. Despite such advancements, many countries do not have tailored policy frameworks today and there still is a large unmet medical need: of the known rare diseases, it is estimated that only 5% have a licensed treatment.

This is due to the fact that a single condition affects relatively few patients worldwide. However globally, the total number of patients impacted by rare diseases is estimated to be 350 million people, approximately 5% of the world population. As such, IFPMA members believe that a shift in mindset is required whereby rare diseases are not considered in isolation but rather regarded as a significant factor within health policy frameworks.

Most rare diseases are genetic

Most rare diseases are genetic and can be associated with life-long disability, often starting in childhood. Many are chronic, degenerative and life-threatening conditions. Many rare disease patients are undiagnosed. A supportive policy environment is therefore necessary to foster greater understanding of these diseases and how they impact patients, to stimulate more research, to encourage appropriate disease management and empower patients and their wider communities.

Our vision is to promote equitable and timely access to the tools and the appropriate healthcare infrastructure and supportive care that patients need to manage their diseases. Our Members believe that each person with a rare disease should expect to be treated with dignity and appropriate care, and we propose guiding principles that would contribute towards the design of a global policy ecosystem that recognises the need to improve the lives of patients with rare diseases.

POLICY PRINCIPLES

Ensuring rare diseases are a public health priority

Addressing the needs of rare disease patients should be a public health priority. Public policy support is crucial to stimulate and accelerate the actions required to improve rare disease care. Patients should have equitable and timely access to the information, services and products they require to manage their disease. A prerequisite to patient access to treatment, including medicines, is access to healthcare professionals and a healthcare infrastructure that fosters the correct diagnosis and can administer appropriate, ongoing disease management.

The International Rare Diseases Research Consortium (IRDiRC) has teamed up researchers and organizations investing in rare diseases research in order to achieve two main objectives by the year 2020 - to deliver 200 new therapies for rare diseases and the means to diagnose most rare diseases.

The road to an enabling environment for rare diseases:

Rare disease should be a public health priority. Globally, the number of patients impacted by rare diseases is estimated to be 350 million people, approximately 5% of the world population. Rare disease patients experience a mix of sensory, motor, mental and physical impairments which could be better addressed by the implementation of appropriate, patient-centered, public policies which incorporate rare disease care into health system design strategies.

Empowering patients and their wider communities

Rare disease patients should be empowered to play a greater role in managing their disease and influencing decisions that affect them. Due to the complexity of rare diseases and their severe debilitating and often life-threatening nature, patients have a unique insight that should be better understood and harnessed.

As the umbrella organisation for rare diseases in Europe, EURORDIS represents patients on committees such as the European Union Committee of Experts on Rare Diseases, the European Medicines Agency scientific committees and working parties and the International Rare Disease Research Consortium (IRDiRC). [1]

The road to patient empowerment:

- **More access to information on their disease and management.** Patient-centred communication is critical to bring the patients closer to the scientific and political communities and inform further research and policy outcomes. Disease awareness and patient-led education initiatives also can help to reduce the misdiagnosis rate, which is often high within rare diseases, and guide patients to the most appropriate treatments.
- **More emphasis on patient-reported outcomes registries.** Patient registries are invaluable in helping to translate basic and clinical research into therapeutic solutions. By complementing clinician-reported data, patient-reported outcomes will also help to improve the robustness, comprehensiveness and quality of registries.
- **Greater involvement in clinical research and evidence-based decision making.** Patients and their caregivers are often very well-educated about their conditions, and can help to bring patient perspectives to the healthcare professionals who they go to for care. Patient networks are therefore often drivers of R&D and when involved early on in clinical trial participation and design, can have a significantly positive impact on drug development.
- **Greater involvement in service configuration for rare disease care.** Patients and their families face numerous challenges in managing their conditions, including, for example, delays in getting diagnosed, finding appropriate specialist providers, or getting help at home or personal care.
- **Enable patients to be partners in regulatory decision making.** Rare disease patients should have a meaningful voice in regulatory and reimbursement assessments.

1. Mavris, M. and Le Cam, Y. (2012), "Involvement of Patient Organizations in Research and Development of Orphan Drugs for Rare Diseases in Europe", Molecular Syndromology, 2012 Nov; 3(5), 237-243 *https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3531929/*

Promoting continued Research and Development (R&D)

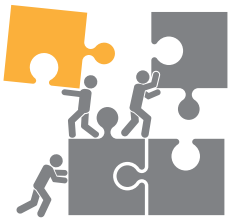
Although considerable progress has been made in the field of rare diseases, the majority of patients suffering from any one of the 7,000 or so rare diseases still have no effective treatment options. An enabling environment, including a supportive regulatory and intellectual property (IP) framework, as well as the commitment of healthcare services to invest in better care for patients with rare diseases, provides the confidence and stability required to nurture investment and stimulate change. International research collaboration efforts across all fields of biomedical research are also key to discovering more treatments for patients with rare diseases.

There are various examples of where the development of orphan drug legislation has helped to promote and support research in the area of rare diseases. Since the introduction of a key regulatory provision in the United States of America in 1983 – the Orphan Drug Act – over 500 orphan drugs have been approved by the US Food and Drug Administration (FDA).[2]

The road to fostering further R&D:

- **Greater collaboration** between profit and not-for profit rare disease researchers. It is not just the diseases which are rare; expertise is as well. International collaboration has an even higher added-value in the case of rare diseases and is of major importance given their specificity – in other words, the limited number of patients and scarcity of expertise. Knowledge must be shared and resources combined as efficiently as possible to tackle rare diseases effectively as a whole.
- **Funding for basic research.** Basic research is crucial for identifying the causes and molecular mechanisms of rare diseases, associated disease targets and the development of diagnostics methods. However, funding for basic research on conditions that affect small numbers of people is often scarce.
- **Regulatory frameworks should enable innovation.** Due to the high unmet need faced by many rare disease patients, orphan drugs are often granted breakthrough therapy designation and faster approvals. Trial feasibility and finding other flexible solutions to the logistical challenges of clinical development should also be considered by regulatory authorities. Rare diseases pose unique challenges to the clinical development process, such as finding a sufficient number of people to participate in clinical trials, or even once they have been identified, addressing other barriers to participation such as frequent travel requirements or overly restrictive inclusion criteria.
- **Early dialogue with regulatory authorities.** Orphan drug development would also benefit from early and continuous dialogue between applicants and regulatory authorities. The ability to react flexibly and in a timely manner to unforeseen challenges during the development process increases the chances of a successful registration and with that the availability of novel drugs for patients with high unmet medical need.
- **Better collection of data and use of disease registries.** Data sharing is particularly important in rare disease research, as small numbers of patients can be barriers to achieving major breakthroughs. Patient registries are critical tools to inform clinical research, to help identify R&D priorities, and orient and optimize rare disease care environments. A registry regularly updated with high-quality data for each rare disease will make it easier to assess the efficacy of a drug in larger numbers of patients and provide valuable feedback on the effects of a drug.

2. PhRMA (2016), "Great Strides Against Rare Diseases"
http://phrma-docs.phrma.org/sites/default/files/pdf/great-strides-against-rare-diseases.pdf



Ensuring sustainable patient access to diagnosis, treatment and care

Fragmented health systems can often lead to patients not being able to access adequate ongoing care and disease management, which remains crucial for people with rare diseases even where no treatment is available. Better integration of the different components of a health system and improved coordination of individual care would enhance the quality of life for people with rare diseases. A multidisciplinary approach to providing care is critical.

Significant strides have been made in some geographies in improving access to specialised care and healthcare provision for patients with rare diseases. For example, nearly all countries in the European Union have now established Centres of Expertise (CEs), which are physical expert structures for the management and care of rare disease patients and which are recognised as a solution to the challenge of providing effective healthcare to rare disease patients.

The road to accessing better care:

- **Greater knowledge and awareness of rare diseases by healthcare practitioners (HCPs).** Physicians (both primary care and specialists) have limited resources and information to properly diagnose and manage patients with rare diseases, compared with more common diseases. At the same time, HCPs have a key role in supporting patients with a rare disease, across the life course. They play a central role in coordinating care and brokering linkages with specialist health and social support services. It is therefore important that they have access to accurate, locally relevant information that can assist them in making the right diagnosis.
- **Screening and better diagnostic testing.** Making diseases easy to diagnose at an early stage can have a significant positive impact on a patient's life, even in the absence of an adequate treatment. Diagnosis in particular represents an important tool in reducing the burden of rare disease. It can take an average of five years for a patient to receive a proper diagnosis. In the meantime, misdiagnosis can have significant impacts on healthcare systems and resources, leading to severe consequences including inappropriate and costly medical interventions such as surgery and psychological treatment.
- **Specialized support services.** Patient access to specialized services is critical. Health systems should be able to integrate support services specific to rare diseases into existing ones, in particular taking account of the consequences and disabilities that rare disease patients suffer due to their conditions.

Where treatment options exist, all stakeholders must work in a collaborative way to find a sustainable way of ensuring patients have access to the treatments they require. Given the different nature and maturity of national healthcare systems, solutions must be tailored. Experience in the treatment of other disease areas has demonstrated that by working in a multistakeholder partnership, solutions can be found to enable patient access.

IFPMA

Learn more and stay connected

www.ifpma.org
Chemin des Mines 9
P.O. Box 195
1211 Geneva 20
Switzerland