

## Response by Rare Diseases International

### Introduction

[Rare Diseases International \(RDI\)](#) welcomes the opportunity to respond to the open consultation on [Draft Concept Note towards WHO's 13th General Programme of Work 2019–2023](#).

RDI is the global alliance of people living with a rare disease of all nationalities across all rare diseases. RDI brings together national and regional rare disease patient organisations from around the world as well as international rare disease-specific federations to create the global alliance of rare disease patients and families.

### Raising the awareness of rare diseases

With more than 300 million people affected, and more than 7000 different types of diseases, most of them genetic and affecting children, rare diseases represent a huge challenge in public health that has been not appropriately addressed yet.

This is a field that would benefit greatly from a globally-concerted action and where the value added of international collaboration is the most visible. **We want with this response reiterate the importance of rare diseases, which we regret was unfortunately not included in the Draft Concept Note.**

**We believe the World Health Organization has a pivotal role to play in addressing the challenges of rare diseases.** There are great inequalities in the field of rare diseases that are not only visible between developed and developing countries but also amongst more economically advanced countries and amongst diseases. By taking on a global leadership role in this area, the World Health Organization has the potential to greatly improve equity of access to health for people with rare diseases across the world, moving the world closer to the achievement of the health-related Sustainable Development Goals.

### The relevance of rare diseases to achieve the Health-related SDGs

The third Sustainable Development Goal underlines everyone's absolute right to live in good health. Yet today, too many people remain left behind, with no access to quality care. Among those who are overlooked are millions of people living with rare diseases. Thus, our community greatly appreciates the focus that the current Draft Concept Note has on helping countries to achieve universal health coverage.

Moreover, a definition of universal health coverage that pays tribute to and adequately values the **importance of diagnostics and preventive services and highlights the need to consider individual determinants of health such as genetics** (page 5) represents an excellent example of a vision aimed at ensuring that a holistic approach to health is undertaken, an approach that goes well beyond the disease itself. The link between this vision and rare diseases is strong and meaningful.

The rare disease patient community also applauds WHO's leadership on health-related SDGs including the most vulnerable population groups with the firm intention to leave no one behind.

With particular reference to **universal health coverage** "including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all" (**SDG target 3.8**), according to our [Joint Declaration](#) in order to

improve the situation of people with rare diseases throughout the world, we believe that attention should focus on:

- Improving **access to services** that will facilitate informed decisions about prevention and screening that are legally permissible, and improve access to accurate and timely diagnoses;
- Facilitate **universal access to high quality, adequate and continued healthcare** and treatments for rare diseases patients, including but not limited to diagnostic tools, surgeries, special diets, transplants, assistive devices, palliative care, psychological support, rehabilitation, access to common and orphan medicinal products;
- Create policies on rare diseases that promote **equity for patients and families without risk of impoverishment**, such as differential approaches, financial protection and specific measures in regional / national / international strategies, including identification and support of specialised expert providers as well as their national and international networking;
- Promote **recognition that rarity requires increased international cooperation and mobility of experts as well as of patients** when expertise is not available locally.

Furthermore, as rare diseases affect primarily children in their first years of their lives, a holistic approach to rare diseases from prenatal screening to adequate care can hugely contribute to the global commitment to **end preventable deaths of newborns and children under 5 years of age** (SDG 3.2) and to **reduce deaths by one-third premature mortality from non-communicable diseases** (SDG 3.4).

### Increase focus of investments in R&D for rare diseases

**Only 5% of rare diseases have treatment options and these are often extremely expensive.**

Innovation is key if we are to meet the challenges posed by rare diseases. This is particularly true in areas where the market does not provide the incentives needed to develop new treatments. The WHO has the authority to initiate and facilitate a global discussion on the development of appropriate funding mechanisms to ensure treatments also reach people in low income country. Too often we fail to appreciate the vital knock-on effects that research on rare and orphan diseases can have on treating more common diseases, serving as a model to help develop effective drugs on a larger scale.

Investing in R&D and thinking about how to improve access for health products for rare diseases is also a way to address many challenges of the future of healthcare for non-communicable diseases and also infectious diseases. We thus commend the focus that the Draft Note puts on fostering innovation. At the same time, we look forward to **making rare diseases visible in health information systems through appropriate coding and seeing an improved classification of rare diseases in ICD11**, to help providing data for clinical research which is critically needed in this field.

### Conclusions

We welcome once again the possibility to comment to the draft concept note with a view to prepare the forthcoming WHO strategy. We stand ready, as rare diseases advocates, to support the WHO in delivering its aims and objectives in the appropriate partnership framework.