



## Response by Rare Diseases International (RDI)

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

### 1. Improving universal health coverage for people with rare diseases

In the same spirit as the definition of UHC indicated in page 7 of the Draft general programme of work: *“UHC means that all people and communities can use the promotive, preventive, curative, rehabilitative and palliative health services that are appropriate to their needs and of sufficient quality to be effective, while also ensuring that the use of these services does not expose the user to financial hardship”*, RDI identifies **access** as one of the key milestones along the journey towards universal health coverage (UHC) and the “1 billion” target and would like to stress that access to affordable and quality-assured health services must be **truly universal** and therefore be achieved for rare diseases patients as well. For this, policies on “quality-assurance of products through effective regulation” and “fair pricing” (page 8) must indeed be pursued, implemented and properly monitored.

In the case of rare diseases, there is a need for further coordination of research and development in therapies focusing on **real unmet needs**. This must be done on the basis of clear data collection and improvement in the development process of therapies, as outlined in the sections below. In particular, in a similar line as “supporting the availability of quality-assured generic products for procurement by global agencies and countries” (page 8), we recommend in particular for the case of rare diseases, exploring and supporting initiatives for joint procurement of medicines and health products to treat people with rare diseases in line with experiences currently ongoing for example in certain regions in Europe.

Rare disease patients and their families are a particularly vulnerable group of citizens who experience scarcity of medical knowledge, difficulties in accessing care and isolation from society due to the rarity of their condition and the scattered expertise. Therefore, considering these distinctive characteristics, we suggest the inclusion of the following mention in page 8, paragraph 3: *“Through its equity and human right lens, WHO sees the health of migrants and refugees as a critical element of UHC and will help countries to address this challenge. More generally, to leave no one behind, efforts in support of UHC must focus on marginalized, stigmatized and hard-to-reach people of all ages, and persons with disabilities **and rare diseases**, in order to ensure that their right to health is translated into reality. Successful progress towards UHC also requires a pro-equity position to be adopted”*.

For this pro-equity position to be properly adopted, the specific needs of this vulnerable population must be considered. As mentioned in page 7 “making health care to be truly universal, calls for a shift from health systems designed around diseases and health institutions towards health systems designed for people.” In this case, we are talking about an estimated 300 million people affected by more than 6000 diseases, a public health priority that cannot be denied. Distinct policies that promote equity for patients with rare diseases and their families without risk of financial hardship



could include support of specialised expert providers as well as differential financial protection policies.

## 2. Improving the lives of people with rare diseases

The “1 billion lives improved” is by all means an important and ambitious objective, but performance should not exclusively be evaluated against numeric criteria. **It is equally crucial to ensure that segments of the population (including people living with rare diseases) see their lives improved regardless and despite the prevalence of their disease.**

We welcome the increased attention given to Non Communicable Diseases and agree that in order to attain the health-related SDGs, NCDs have to remain in the list of priority areas. However, the overall target will not be attained if most of the resources are allocated to reduce environmental risk factors alone. Rare, genetic and inherited diseases should be part of the NCD agenda.

In a similar way, the justified target on women, children and adolescents will fall short if it does not include attention to rare congenital disorders. Moreover, 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 by one-third premature mortality from non-communicable diseases (SDG 3.4).

It would be worth including in the Impact Framework (page 3 – target ‘Reduce the rate of under-five child deaths by 30%’) **a specific goal to attain an adequate level of newborn and pre-natal screening for rare diseases to ensure adequate care from birth** and avoid potentially complications thus improving maternal health. The WHO’s role could be to promote the uptake of appropriate guidelines on pre-natal and newborn screening worldwide according to international standards.

## 3. Improving data collection on rare diseases

While sharing of health data is supported by the majority of patients, those living with a rare disease are particularly in favour. Information is typically scarce for each individual rare disease. **The ability to share and pool data is essential in the rare diseases field and in all fields requiring a specific concentration of expertise: only through data congregation can one attain a critical mass, which generates knowledge and drives forwards improvements in research and healthcare.**

Considering that the WHO will “focus on roles where it has a comparative advantage” (page 18) and in “filling critical data gaps” (page 19), it is critical to focus investments in data collection for rare diseases as there is a clear gap that needs to be filled which will contribute to the achievement of UHC and health SDGs, in particular but not limited to the reduction of premature mortality of children under 5.

Rare diseases are an area that desperately needs standardisation of data collection to improve epidemiology, understanding of the diseases, diagnosis and efficient care to improve outcomes. It is particularly problematic that there is a lack of systematic use of recognized and widely used codes



and computer readable ontologies able to classify and code the rare disease encountered and a comprehensive description of its symptoms thus enabling the identification and diagnosis of a patient's disease.

In addition, it is also necessary to overcome the heterogeneity of electronic health records (if existing). The collection of data in a standardised manner would make it interoperable and enhance its power making it possible to exploit the potential given by advancements in IT technologies for health purposes like for instance high-performance computing of genetic information which is crucial to accelerate diagnosis. The wealth of unstructured data (from social media, apps, wearables) is exponentially increasing making big data analytics crucial in therapy development, treatment and disease monitoring.

Therefore, in addition to “maintaining the International Classification of Diseases” (page 18), it would be necessary to **update it**, particularly considering its 11<sup>th</sup> revision, (ICD-11), to include appropriate coding and improved classification of rare diseases that will help provide data for clinical research which is critically needed in this field.

#### 4. Fostering innovation in 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. It is crucial that innovation and R&D processes deliver outputs that are truly meaningful to those who are meant to benefit from innovative services and products. Ensuring that innovation is not simply and exclusively market-driven but rather shaped on the primary needs of all patient populations (including people with rare diseases, migrants and refugees, low income countries) will represent a marked advancement towards sustainable and patient-centric UHC and is a role that the WHO should particularly embrace as “identifier and connector” (page 20).

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.

**The WHO can become the innovation catalyst for creating new ways to make therapies development faster and at lower cost.** Several tools, techniques or methodologies exist today that can allow medicines to come to market in greater numbers and for lower investments. There is good reason to believe that they would dramatically reduce the cost of R&D for rare disease therapies, reduce the number of patients who need to be involved in clinical research and also reduce the overall time necessary for full product to come to the market.