



Rare Diseases: The Missing Keystone of Universal Health Coverage

Executive Summary

UN General Assembly Resolution 72/139 from 12 December 2017 on Global Health and Foreign Policy marked a milestone moment in the history of global health policy. Not only did the resolution highlight the need to address the health of the most vulnerable for an inclusive society, thus underlining that the health of the most vulnerable is both a core component and a condition of an inclusive society; but it also spelled out the importance of placing "a specific emphasis on the [...] vulnerable and marginalised segments of the population" at the heart of any strategy to progress towards the goal of universal health coverage.

Persons living with rare diseases count amongst the most vulnerable and marginalised populations on the planet today. Rare diseases are a family of well over 6,000 conditions, often of genetic origin, which affect small to ultra-small populations of patients (typically fewer than 1 in 2,000 individuals). Most rare diseases are complex, chronic, progressive, degenerative, disabling and frequently life-threatening. They lead to serious unmet needs for the persons affected, their families and their caregivers, ranging all across the spectrum from health and quality of life to equality, social inclusion, access to education, but also access to employment and other opportunities to give back to society and be an active member of it – thus encompassing many different dimensions of individuals' fundamental human rights.

When considered as a whole, rare diseases affect an estimated 4 percent of the world's population, thus about 300 million people worldwide at the very least – a figure on par with other major non-communicable diseases. Despite this, rare diseases are all too often neglected, to a large extent because of insufficient medical knowledge about many of them. This lack of knowledge means that many rare diseases are not diagnosed, therefore disabilities and other symptoms are not recognised, and in turn persons with rare diseases do not receive the same support (be it financial, educational or social) as those with more common and well-known disorders. Ultimately, this effectively bars persons with rare diseases from genuine integration into society and, consequently, from achieving the "realization [...] of the economic, social and cultural rights indispensable for [their] dignity and the free development of [their] personality" (art. 22 of the December 1948 Universal Declaration of Human Rights).



Unfortunately, it is a fact that until recently rare diseases have largely been left out thus far from the momentous political conversation on universal health coverage, even though the essence of the aspirations and objectives behind universal health coverage resonate so strongly with the daily experience, needs and expectations of all persons living with a rare disease across the world.

The ambition of this paper is therefore to reconcile for the first time these two fields by offering a reasoned interpretation of why and how rare diseases integrally belong, by their very nature, to the concept of universal health coverage.

We, as the global rare diseases movement borne out of civil society more than 40 years ago, are calling for more than just the “inclusion” or “addition” of rare diseases to the prevailing acceptation of universal health coverage as yet another avenue for action amongst many others. Indeed, we affirm our view that:

- The global rare diseases and universal health coverage movements share a profound commonality of spirit and vision, deeply entrenched in the guiding principles and objectives that sustain their respective aspirations, and in the direct continuity of the aspirations set out in the UN Convention on the Rights of the Child (1989) and the UN Convention on the Rights of Persons with Disabilities (2006). The universality principle, the emphasis on individuals receiving the right health services without hardship, the strive towards a holistic and integrated approach to health, or still the permanent focus on “more than just health” and on the human rights, equity, solidarity and social justice dimensions – all represent living bridges fostering a genuine kinship between the ideals of the global rare diseases movement and the WHO’s ambitions for universal health coverage.
- Universal health coverage shall never be fully attained nor realised if persons living with rare diseases are left behind and their needs are not met. The major theories of social justice forming the warp and weft of universal health coverage all recognise the importance of a substantial focus on the most vulnerable and neglected communities in society. Furthermore, there is ample research and literature to underline how, even in resource-constrained settings, it is perfectly possible – and actually in scope – to prioritise rare diseases even in the earliest stages of planning for universal health coverage. This ultimately remains a political responsibility, a governance decision and a societal choice rather than a purely economic one, and the implications of this must be fully acknowledged if the transformative potential of universal health coverage is not to be undermined. Nevertheless, a number of recent developments across the world offer hope and reassurance that national governments and regional organisations are increasingly not only understanding the structural importance of rare diseases in a universal health coverage context, but also reflecting it in practice in new policies and legislation.
- Now is the time for integration of rare diseases in the political declaration on universal health coverage. Throughout the world, civil society is, and will continue to be, a leading partner to catalyse the transition of existing healthcare systems towards universal health coverage models.



The global rare diseases movement is ready today to take its rightful place amongst other components of international civil society to support and accomplish that ambition, and we have over the past few years started to develop first tangible proposals to that end. Across the world, our community has decades of medical knowledge, patient education campaigns and a wealth of other resources that are all waiting to be put to good use to make universal health coverage not just a reality, but a success.

The global rare diseases movement looks forward to contributing to the Political Declaration on Universal Health Coverage and to being invited and represented at the 23 September 2019 UN High-Level Meeting on Universal Health Coverage in New York.

We are impatient to demonstrate our readiness to « move together to build a healthier world », one in which the 300 million people living with a rare disease worldwide will feel at long last more widely recognised, better supported and fully associated to one of the most inspiring endeavours of this century.

We hereby call on all partners – UN agencies, NGOs but also the Group of Friends of UHC and all other national governments – to join us.
