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 acception 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.

Foreword: Firming up the contours of universal health coverage

Since 2005 and its first ever appearance in an official political statement from the United Nations[1] [2], the concept of universal health coverage has become "*an increasingly central rallying point for global health advocacy*"[3]. In parallel, the concept has also undergone over the years a number of evolutions and refinements – not least to limit the profusion of diverse and sometimes disconnected definitions and, in turn, to help bring greater unity to its denomination, scope and ambition[4].

For the present paper, we decided to take as a central point of reference the latest definition proposed and publicised by the World Health Organisation itself, i.e.:

> *« Universal health coverage means that **all individuals and communities receive the health services they need without suffering financial hardship**.*
>
> *It includes the full spectrum of essential, quality health services, **from health promotion to prevention, treatment, rehabilitation, and palliative care.***
>
> ***Universal health coverage enables everyone to access the services that address the most important causes of disease and death,** and ensures that the quality of those services is good enough to improve the health of the people who receive them.*
>
> *[…] Universal health coverage is not just about health financing. **It encompasses all components of the health system:** health service delivery systems, the health workforce, health facilities and communications networks, health technologies, information systems, quality assurance mechanisms, and governance and legislation.*
>
> *Universal health coverage is not only about ensuring a minimum package of health services, but also about ensuring a progressive expansion of coverage of health services and financial protection as more resources become available.*
>
> *[…] Universal health coverage is comprised of **much more than just health**; taking steps towards universal health coverage means steps towards **equity, development priorities, and social inclusion and cohesion**. »*[5]

[1] World Health Assembly Resolution WHA58.33 on "*Sustainable health financing, universal coverage and social health insurance*", May 2005 Accessible here > bit.ly/2E2eRFS

[2] While we refer herein to the May 2005 World Health Assembly Resolution as a milestone political statement and a generally acknowledged starting point for the contemporary concept of universal health coverage, it is only fair to also note that the concept can actually be traced back much earlier in time. It is for instance present in germ form in paragraph 6 of the final declaration of the September 1978 International Conference on Primary Health Care in Alma-Ata, USSR, which reads: *"primary health care is essential health care […] made universally accessible to individuals and families in the community […] at a cost that the community and country can afford"*. See here > bit.ly/2G4IQQ6 The aspirations and commitments of the 1978 Alma-Ata Declaration were most recently reaffirmed at the Global Conference on Primary Health Care in October 2018 in Astana and in the ensuing Declaration, accessible here > https://bit.ly/2ORMhNZ

[3] "*The evolution of the field of Health Policy and Systems Research and outstanding challenges*", Bennett et al. Health Research Policy and Systems (2018) 16:43. Accessible here > bit.ly/2E3dOFE

[4] "*What does universal health coverage mean?*", O'Connell et al. Lancet (2014); 383: 277–79. Accessible here > bit.ly/2RHQ0LH

[5] "*Universal health coverage (UHC)*", WHO Fact Sheet, last updated December 2018. Accessible here > bit.ly/2O22lsl

1. The Global Rare Diseases Movement and Universal Health Coverage: A Commonality of Spirit and Vision

Commented [A1]: Following a comment by Amanda Bok (European Haemophilia Federation), we are tempted to complete this one section by also drawing on a longer historical perspective and on the principles enshrined in the UN Convention on the Rights of the Child (1989) and that on the Rights of Persons with Disabilities (2006).

To be further developed…

DRAFT – A brief executive summary may yet be added at the start of each section.

Rare diseases were coined as a specific issue deserving social interest less than 35 years ago and in different parts of the world simultaneously, from the United States all the way across to Europe and Asia. In each region, representatives of what were very small patient organisations at the time came together with pioneering doctors and geneticists to begin to form a better understanding of the particular difficulties associated with the rarity of a given disease, but also of the conditions required for a broader societal effort towards overcoming these difficulties.[6]

"If we are serious about leaving no one behind, then we cannot leave behind people with rare diseases just because they are few."

Nata Menabde, WHO,
11 November 2016

Over time, the rare diseases movement structured itself more decisively – first at the national level, then at the regional level and most recently at the global level. Annual observances like Rare Disease Day[7] (initiated in 2008) but also platforms like the International Rare Diseases Research Consortium[8] (IRDiRC, founded 2011), Rare Diseases International[9] (RDI, founded 2014) or still the NGO Committee for Rare Diseases at the United Nations[10] (founded 2016) have all played a seminal role to make the voices, needs and expectations of the 300+ million people living with a rare disease worldwide better heard and understood within the UN system but also in the broader concert of nations at large.

Along that journey and at the initiative of each of these platforms, a number of milestone declarations and political statements were adopted by the global rare diseases movement to affirm its identity, beliefs and aspirations. The Yukiwariso Declaration launched by ICORD[11] in Tokyo in 2012, the Joint Declaration on "*Rare Diseases: An International Public Health Priority*" launched by RDI in Madrid in 2015 or still the Founding Act of the NGO Committee for Rare Diseases launched in New York in 2016 all converged to call for a more resolute and better coordinated international collaboration to address the challenges posed by rare diseases.

[6] "*How It All Started: The Contribution of the NGO Community to Advancing the Causes of Rare Diseases*", speech by Mr Terkel Andersen, EURORDIS-Rare Diseases Europe, at the Global Gathering for Rare Diseases in New York, 11 November 2016
Accessible here > bit.ly/2QjYCLT
[7] https://bit.ly/1d4B51n
[8] https://bit.ly/2vFwUhs
[9] www.rarediseasesinternational.org
[10] www.ngocommitteerarediseases.org
[11] www.icord.se

This trajectory towards heightened global awareness is not fundamentally different in essence from that which saw the concept of universal health coverage come (over many more years, however) to the prominence it enjoys today in international policy circles.

But the most striking similarity between the global rare disease movement and the universal health coverage one is not one of direction or itinerary – **rather one of substance**.

This similarity appears more clearly through a cross-comparison of the core principles at the heart of the concept of universal health coverage on the one hand, and of the central tenets and messages contained in each of the major statements on rare diseases referred to hereabove. Table 1 below illustrates such a comparison.

WHO: Principles of Universal Health Coverage (as per the December 2018 WHO Fact Sheet)[12]	**ICORD: Yukiwariso Declaration** (2011-2012)[13]	**RDI: Joint Declaration** (2015)[14]	**RDI: International Recommen-dations to Address Specific Needs of Undiagnosed Rare Disease Patients** (2016)[15]	**NGO Committee for Rare Diseases: Founding Act** (2016)[16]
Universality: *"... all individuals and communities receive the health services they need..."*	*"... the ethical principle of justice requires that the needs of rare disease populations are specifically addressed, as they should be for any minority or underserved community..."* (pg. 4) *"... [the] rarity of a disease should not be grounds for denying access to services or therapies..."* (pg. 6)	*"3. Support and empower patients and families to [shape] national health care provision that is appropriate to their needs"* *"7. Facilitate universal access to high quality healthcare and treatments for rare diseases patients [...]"*	*"... undiagnosed rare disease patients should be recognised as a distinct population with specific unmet needs by national authorities to enable development of personalised health and social care..."* (pg. 1)	*"... no one country, no one continent alone can solve the problems posed by rare diseases..."*
Financial Protection: *"... without financial hardship... reduc[ing] the risk that people will be pushed into poverty"*	*"... rare diseases create disparities and vulnerabilities in health status for affected populations and [governments] should put in place specific policies to address them... "* (pg. 6)	*"12. Elaborate policies based on common values (equity, solidarity and social justice) that have a positive impact on the lives of rare diseases patients"*	*"... in most countries, undiagnosed rare disease patients remain an invisible and highly vulnerable population..."* (pg. 4) *"... late diagnoses [...] can have irreversible and life-threatening consequences; undiagnosed rare disease patients and families also face added social and daily life challenges..."* (pg. 3)	

[12] See footnote 4
[13] bit.ly/2G1NWfT
[14] bit.ly/2QfXA3q
[15] bit.ly/2BSVX2y
[16] bit.ly/2L2OKAg

WHO: **Principles of Universal Health Coverage** (as per the December 2018 WHO Fact Sheet)[12]	**ICORD:** **Yukiwariso Declaration** (2011-2012)[13]	**RDI:** **Joint Declaration** (2015)[14]	**RDI:** **International Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients** (2016)[15]	**NGO Committee for Rare Diseases:** **Founding Act** (2016)[16]
Holistic Approach to Health: *"... the full spectrum of essential, quality health services, from health promotion to prevention, treatment, rehabilitation, and palliative care..."*	*"... governments should adopt policies that aim to achieve equitable allocation of resources towards all aspects of rare diseases, incl. research, clinical care, information resources and development of treatments..."* (pg. 6)	*"1. Enhance visibility of rare diseases [...] in terms of public awareness and within the healthcare system [as] a public health priority"* *"6. Improve access to services that will facilitate informed decisions about prevention and screening [...] and improve access to accurate and timely diagnosis"*		*"... rare diseases fit within the objectives of several UN SDGs (1, 3, 4, 5, 8, 10 and 17) and can significantly contribute to their achievement..."*
Quality of Care: *"... ensur[ing] that the quality of those services is good enough to improve the health of the people who receive them..."*	*"... responses such as prioritization and the need to ration resources, as reasons for lesser attention to rare diseases [...] are not ethically sustainable arguments..."* (pg. 5)	*"7. Facilitate universal access to high quality healthcare and treatments for rare diseases patients [...]"*	*"... to improve outcomes for the 'not yet diagnosed' group, both the route to, and the quality of, diagnostic tools and also access to extensive genomic data need to be improved..."* (pg. 2)	
Integrated Healthcare System Thinking: *"... not just about health financing. It encompasses all components of the health system: health service delivery systems, the health workforce, health facilities and communications networks, health technologies, information systems, quality assurance mechanisms, and governance and legislation..."*	*"... governments should adopt policies that aim to achieve equitable allocation of resources towards all aspects of rare diseases, incl. research, clinical care, information resources and development of treatments..."* (pg. 6)	*"1. Enhance visibility of rare diseases [...] in terms of public awareness and within the healthcare system [as] a public health priority"* *"2. Better classification and codification of rare diseases..."* *"8. Create policies [...] that promote specific measures in regional / national / international strategies, incl. identification and support of specialised expert providers..."* *"11. Build opportunities for effective networking of patient registries"*	*"... undiagnosed rare disease patients require the availability of a complete health and social care pathway in advance of receiving a diagnosis..."* (pg. 2) *"... integrated clinical and social pathways specifically designed to answer [the] needs [of undiagnosed rare disease patients]..."* (pg. 4)	*"... common national policies and international collaboration can address these challenges..."*
Dynamic Approach Over Time: *"... not only about ensuring a*	*"... responses such as prioritization and the need to ration resources,*	*"6. Improve access to services that will facilitate*		

WHO: Principles of Universal Health Coverage (as per the December 2018 WHO Fact Sheet)[12]	**ICORD: Yukiwariso Declaration** (2011-2012)[13]	**RDI: Joint Declaration** (2015)[14]	**RDI: International Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients** (2016)[15]	**NGO Committee for Rare Diseases: Founding Act** (2016)[16]
minimum package of health services, but also about ensuring a progressive expansion of coverage of health services and financial protection as more resources become available…"	*as reasons for lesser attention to rare diseases […] are not ethically sustainable arguments…"* (pg. 5)	*informed decisions about prevention and screening […] and improve access to accurate and timely diagnosis"*		
More Than Just Health: *"… taking steps towards UHC means steps towards equity, development priorities, and social inclusion and cohesion…"*	*"health care and treatment for rare diseases is a human rights issue; non-discrimination, justice and equity of access to health care, all require that specific policies are put in place to address the needs of people affected by rare diseases…"* (pg. 4) *"… a comprehensive approach to rare diseases is needed, incl. education, research, prevention, diagnosis, care and treatment, social support and inclusion…"* (pg. 5)	*"12. Elaborate policies based on common values (equity, solidarity and social justice) that have a positive impact on the lives of rare diseases patients"*	*"… national sustainable programmes dedicated specifically for undiagnosed diseases should be developed […] to enable rapid and equitable access to diagnosis and social support…"* (pg. 1) *"… undiagnosed rare disease patients and families also face added social and daily life challenges…"* (pg. 3) *"… addressing the issues associated with access to social services, education, occupation, and rehabilitation support…"* (pg. 3)	*"… persons living with rare diseases worldwide are facing common challenges across diseases and across borders that affect all parts of their lives…"* *"… rare diseases fit within the objectives of several UN SDGs (1, 3, 4, 5, 8, 10 and 17) and can significantly contribute to their achievement…"*

Figure 2 · Comparing the core principles at the heart of the concept of universal health coverage with central tenets and messages from major rare disease statements and declarations (2011-2016)

This succinct comparison highlights several areas of meaningful convergence:

- The **universality** principle whereby all individuals and communities should receive the health services they need is inseparable from the views upheld by the global rare diseases movement, which contends that people living with a rare disease (and even more so, undiagnosed rare disease patients) should be recognised as a distinct community with very specific needs to be met.
- The emphasis placed on **receiving the right health services without hardship** resonates with the general recognition that people living with a rare disease are highly vulnerable and at a greater disadvantage compared to the general population, and that they therefore need specific support underpinned by the principles of equity, solidarity and social justice to protect them from social challenges and poverty. The high level of medical complexity inherent to rare and ultra-rare

diseases also goes hand in hand with a greater need to make high-quality healthcare accessible to affected individuals and their families, fully in keeping with the spirit of universal health coverage.

- All of the major statements on rare diseases listed hereabove promote **a holistic and integrated approach to health**, ranging comprehensively all the way from governance to system design, from resource allocation to actual care delivery, or from research to treatment. In that regard, they are well aligned with the currently accepted concept of universal health coverage and supportive of the aspiration not to reduce UHC to health financing merely.
- Above all, the understanding that **universal health coverage is "more than just health"** is sharply mirrored in the views advocated by the global rare diseases movement, which has repeatedly positioned healthcare for rare diseases as not only a health issue but even more fundamentally one of human rights, equity, solidarity and social justice.

As a result, it emerges as a reasonable and legitimate conclusion to draw that the core principles, messages and demands that have been at the cornerstone of the global rare diseases movement's advocacy and action over the past half-decade bear not just a substantial degree of similarity – *but, even more so, a genuine commonality* – with those heralded by the universal health coverage movement.

2. No Universal Health Coverage Without Rare Diseases:
An Ethical Imperative Anchored in the Theories of Justice – and a Political Choice

DRAFT – A brief executive summary may yet be added at the start of each section.

Although essential, underlining the kindred spirit and shared fundamentals between the universal health coverage and rare diseases movements remains far off the mark. The collective reflection on the place of rare diseases in the current and future debate on universal health coverage shall not be complete nor relevant if **the impossibility to fully achieve universal health coverage without a strong emphasis on rare diseases** is not properly understood and, in turn, acted upon by policymakers.

This was the exact meaning of the address Helen Clark, former Administrator of the UN Development Programme (UNDP), delivered to the 2016 ICORD Annual Conference in Cape Town, as she noted that "*no country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases*".[17]

"No country can claim to have achieved universal healthcare if it has not adequately and equitably met the needs of those with rare diseases."

Helen Clark, UNDP, 20 October 2016

That statement was naturally in line not only with the content of Sustainable Development Goal no. 3, "*ensure healthy lives and promote well-being for all at all ages*", target 3.8 of which aims to "*achieve 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*" but also, reaching even farther back in time, with the ambition of "*achieving health for all*" expressed at Alma Ata in 1978.

But, from a broader perspective, **the notion that meeting the needs of people living with rare diseases is an essential condition for achieving universal health coverage – and, simply, a moral imperative – takes its roots much more deeply into major contributions to 20th century political philosophy**.

For instance, the question of the level of recognition and attention to be granted by a society or healthcare system to rare diseases finds a number of answers in the eminent theory of justice developed by John Rawls[18], two fundamental principles of which are:

[17] https://bit.ly/2gJ9bWG

[18] As very well captured in the 2011-2012 ICORD Yukiwariso Declaration bit.ly/2G1NWfT. The theory of justice was first outlined by John Rawls and can be accessed in the following publications. John Rawls (1971) A Theory of Justice. (Cambridge: Harvard University Press). 624pp and John Rawls (2001) Justice as Fairness: a Restatement. Ed. Erin Kelly (Cambridge, M.A.: Harvard University Press) 214pp.

1. Each person is to have an equal right to the most extensive total system of equal basic liberties compatible with a similar system of liberty for all.

2. Social and economic inequalities are to be arranged so that they are both: (a) to the greatest benefit of the least advantaged, consistent with the just savings principle; and (b) attached to offices and positions open to all under conditions of fair equality of opportunity.[19]

In a 2014 article exploring the implications of Rawls' theory of justice on funding for orphan medicines,[20] Hyry, Roos and Cox also concluded that "*the concept of social justice outlined by Rawls shows that a distinction between the individual and society [...] is bogus*". Their analysis goes on to dispel the fallacies that a society could not rationally choose to prioritise and fund a life-saving, expensive treatment for a very rare disease, or – more broadly – that the interests of a minority of individual patients should necessarily and by default enter into conflict with the aims of public health and with the best interests of society at large. The article also affirms as a corollary and direct consequence of Rawls' thinking that "*those lucky not to suffer from the effects of a rare disease should not use their morally arbitrary good fortune to disadvantage those who do*" and that, in a genuine Rawlsian model, everyone must have an equal right to pursue their life plans, to the extent that ***"the more a disease impairs a person's capacity to pursue their goals, the more urgent it is that their health need is addressed"***.

"Rare disease patients are a particularly vulnerable group of citizens who experience scarcity of medical knowledge, difficulties in accessing adequate care, as well as isolation from society due to the rarity of their condition and the scattered expertise."

Rüdiger Krech, WHO,
11 May 2018

This emphasis on the equal right of all individuals to pursue their life aspirations regardless of their condition or health status also resonates with the political discourse on social justice put forward in recent decades by Amartya Sen, and particularly with his approach centered around the concept of capability (defined as "*the ability to satisfy certain elementary and crucially important functionings up to certain levels*"[21]). In The Idea of Justice (2009), Sen himself notes that ***"people with physical or mental disability are not only among the most deprived human beings in the world, they are also, frequently enough, the most neglected"***.[22]

Elsewhere[23], Sen remarks how that neglect from the main schools of thought in theories of justice has tended in turn "*to bias practical policies in the direction of inaction, [even contributing] to suppressing the sense of inadequacy that can reasonably accompany the failure to take* ***a responsible view of the social***

[19] John Robert Wilson Jr. "*Ethics in the service of the rare disease patient: Application of the thought of John Rawls and Paul Ramsey toward the increased availability of orphan drugs*" Master's Thesis, Rice University, 1990 https://bit.ly/2RWDt6W
[20] https://bit.ly/2LelyWN
[21] **Sen** 1992: 45 n. 19
[22] Sen, A. The Idea of Justice, Chapter 12 "Capabilities and resources", section "Disability, resources and capabilities" p. 258 AND ALSO in *Disability and Justice, Keynote Speech by Amartya Sen "Disability and Inclusive Development: Sharing, Learning and Building Alliances", a conference organized by the World Bank, Washington, November 30-December 1, 2004* https://bit.ly/2Ey3EOc
[23] *Disability and Justice, Keynote Speech by Amartya Sen "Disability and Inclusive Development: Sharing, Learning and Building Alliances", a conference organized by the World Bank, Washington, November 30-December 1, 2004* https://bit.ly/2Ey3EOc

obligation to the disabled" – even though "***the tragic consequences of disability can be substantially overcome with determined societal help*** *and imaginative intervention*".

In a 2014 review on the "ethics of the social determinants of health"[24], Jennifer Prah Ruger dwells on a few central tenets of Sen's approach, e.g. the consideration of capabilities as the real freedoms that people have and may enjoy, but also the view that **the expansion of human capabilities, and by extrapolation the expansion of the freedom of individuals to choose the life they want to live, is "*the ultimate end of public policy*".** Even more than Rawls, Sen's thinking values health intrinsically and more directly, and therefore **health-related capabilities such as the avoidance of preventable morbidity or premature mortality are regarded not only as ends in themselves but also as instrumentally important for the achievement of all other ends** (e.g. the degree to which individuals have the capability to participate actively in their work, social, and political life, to be well-educated or to be secure in their economic facilities). In Ruger's words, *"**such social determinants of health have both constitutive and instrumental value**, and they serve not only to contribute to the general capability of a person to live more freely, but they also serve to complement one another*".

The principles set out in the theories of justice of Rawls and Sen – a few of which only were touched upon in the preceding paragraphs – all converge towards a political approach characterised by: an equal right of all individuals to enjoy the full extent of their social freedoms, regardless of the severity of their handicap; a marked commitment and greater sense of urgency in favour of lending more support to those with the most severe handicap, rather than addressing first and foremost the most common needs of the multitude; and even, as in the case of Sen, a view that the ultimate horizon of public policy should be to empower all individuals to enjoy their capabilities to the fullest.

These very principles clearly articulate an inescapable requirement for the needs of people living with rare diseases to be specifically addressed, as they should for any minority or under-served community, especially in the context of a policy of universal scope and ambition such as universal health coverage. Far from being unattainable ideals, these principles are being increasingly recognised and recently found a translation into actual policy at the highest level globally: the Resolution on global health and foreign policy adopted by the United Nations' General Assembly on 12 December 2017 said exactly that: the true benchmark of how inclusive a society can pretend to be is how it addresses the health of its most vulnerable.[25]

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A rapid scan of the existing literature reveals a very dense body of articles and reflections written over the years that approach the question of how to implement universal health coverage chiefly under the angle of **setting priorities to distribute and allocate finite resources or health services as "fairly" as possible**. This

[24] https://bit.ly/2RVqORP
[25] https://bit.ly/2RRyo34

line of reasoning raises a new and no less important question: whilst one may agree to the moral or philosophical imperative of including rare diseases as a major pillar of a true universal health coverage system, **is it realistic, feasible or even fair to do so in a context of often extremely constrained resources and inputs?**

Norheim in 2016[26] agreed for instance that, while the WHO's aspiration to universal health coverage was "*radical*", in the face of evident resource constraints, "*essential health services [could not] entail all possible services but rather a comprehensive range of key services that are well-aligned with other social goals*", and thus that priority-setting was unavoidable on the path to universal health coverage. **Do care and/or therapies for rare diseases fit within these "key services"? The answer offered by Norheim is actually positive.** While the author notes that the size of the population affected by a given need tends to be a contested criterion, he nevertheless concludes that all health benefits are ethically relevant and that "*the ethical importance of services for rare [diseases] is captured by the three accepted criteria*", two of which are "*priority to the worse-off*" and "*financial risk protection*" – both of particular relevance in light of the abundant literature underscoring the often drastic loss of social and economic opportunities incurred by people living with rare diseases.[27]

Persons with rare diseases are often trapped in a vicious cycle of **vulnerability** and **poverty** due to **exclusion** from healthcare and education systems, as well as the job market.

Persons with rare diseases and disabilities are often **marginalised from the job market** not only due to prejudice, stigma and discrimination, but also due to **lack of accessible facilities**, of **flexible working hours** and of adapted roles[28].

50% of all rare diseases affect children, who often face great difficulty to attend school due to **inaccessibility** of facilities and non-adapted teaching methods, but also due to prejudice, stigma and discrimination.

Persons with rare diseases remain **a marginalised and invisible, yet statistically significant population** of nearly 300 million individuals worldwide, suffering from **discrimination** in the areas of health, labour and political inclusion – all prime examples of concrete inequalities that must be addressed in the spirit of SDG 10.

The life-threatening urgency placed on persons with rare diseases to take part in research and clinical trials may also occasionally compromise **their right to the privacy** and due protection of their personal data and health records.[29]

[26] https://bit.ly/2GhAy7C

[27] See for instance Schieppati A, Henter JI, Daina E, Aperia A. Why rare diseases are an important medical and social issue. Lancet 2008; 371:2039–41.

[28] See "*La integración laboral y social de colectivos especialmente vulnerables: personas con enfermedades raras, personas con enfermedades mentales y mujeres con discapacidad*" [online] By María del Carmen López Aniorte, Profesora Titular de Derecho del Trabajo y de la Seguridad Social at Universidad de Murcia, and Gema Chicano Saura, Profesora Asociada de Derecho del Trabajo. Available at: www.ilo.org/madrid/fow/trabajo-decente-para-todos/WCMS_548585/lang--es/index.htm [Accessed 8 Feb 2018].

[29] See "*Universal Declaration on Bioethics and Human Rights*" [online] UNESCO. Available at: portal.unesco.org/en/ev.php-URL_ID=31058&URL_DO=DO_TOPIC&URL_SECTION=201.html [Accessed 8 Feb. 2018].

The responsibility of caring for persons with rare diseases **disproportionately falls upon women, and particularly mothers** as rare diseases predominantly affect young children. Their time and efforts to provide unpaid care and domestic work, as well as **the burden it places on their own careers and pursuits** (going as far as forcing them to abandon their employment, with very direct implications on their income, pension and future social payments as well as **their economic independence** overall), is often undervalued and unrecognised.[30]

Figure 3 · A brief overview, across SDGs, of the multifaceted challenges and risks of loss of social and economic opportunities faced by people living with rare diseases. *Adapted from « The "Right to Health" in Rare Diseases: A Practical Contribution to Implementing and Achieving the Sustainable Development Goals » (NGO Committee for Rare Diseases, Submission to the Office of the UN High Commissioner for Human Rights, February 2018)* [31]

The WHO itself, in its 2014 consultative report Making fair choices on the path to universal health coverage[32], accepts that "*when universal health coverage cannot be realised immediately, making progress fairly and equitably becomes imperative*" and also notes that both fairness and equity "*bear on the critical choices on the path to that goal*". In practical terms, when it comes to covering a larger population under a nascent universal health coverage programme or strategy, the WHO recommends to country authorities to first expand coverage "***for low-income groups, […] and other groups disadvantaged in terms of service coverage, health, or both***" (all populations which very often intersect with people living with rare diseases, as per footnote 24) and goes as far as noting that "*an exclusive focus on cost-effectiveness [concerned solely with the total number of healthy life years] is generally found indefensible […] there are good reasons to start with those worse off over their lifetime*". **All of these elements are highly compatible with a prioritisation of rare diseases even in the earliest stages of planning for universal health coverage.**

More recently still in 2018, in "Health systems implications of rare genetic conditions in low- and middle-income countries: a case study approach"[33], Allotey and Reidpath go one step further and contend that ***"a resilient and responsive health system providing universal health coverage is one that is able to cope with both the commonplace conditions faced by the majority as well as rare conditions, particularly when experienced by more marginalised groups"***. Building on the experience from a real-life case study – a 4-year old child suffering from Harlequin Ichtyosis in Southeast Asia – the authors shine a new and different light on rare to ultra-rare diseases, seen not as a threat to the sustainability of a universal health coverage system, but rather as "***an important litmus test of [its] robustness***".

[30] See "*Juggling care and daily life: The balancing act of the rare disease community*" [online] EURORDIS-Rare Diseases Europe. Available at: www.eurordis.org/news/3000-rare-disease-patients-carers-voice-difficulties-balancing-care-life [Accessed 8 Feb. 2018].

[31] https://www.ngocommitteerarediseases.org/wp-content/uploads/2018/05/NGO-CfRDs-Submission-The-Right-to-Health-in-Rare-Diseases_Feb-15-2018.pdf

[32] https://bit.ly/2S1gjfU

[33] https://bit.ly/2S09oDG

To sum up, there is growing evidence across the literature to affirm that, even within a finite pool of resources, it is not only possible but actually pertinent and mission-critical to prioritise rare diseases upfront as part of the implementation of a universal health coverage system.

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While exploring the same issue as Norheim, Allotey and others, Rumbold, Baker, Ferraz et al. took in 2017 a slightly different route, focusing rather on the perceived frontal opposition between priority-setting in healthcare systems and individuals' right to health.[34] If their main conclusion, like hereabove, is that both notions do actually have a common grounding and can be mutually dependent and mutually reinforcing, they also raise an additional and interesting question by noting that, "*when an acceptable interpretation of the content of the right to health under national law has been clarified, respecting the principles discussed above, **finance ministers should reappraise their budgets, considering the state's obligations under that right. The right to health, just as civil and political rights, requires resources**, whether through taxation or other means. As with civil and political rights, the right to health is supposed to be binding. **When the status quo fails to uphold rights, changes, including judicial remedies, are needed**.*"

This observation highlights an important truth. The decision to implement a universal health coverage system is not, and cannot be reduced to, a purely administrative one that would look at the reorganisation or redistribution of a finite pool of resources across many competing targets and objectives. **It is by essence a political responsibility, a governance decision and a societal choice,** which should dare question upfront whether the size of the existing pool of resources is adequate and commensurate to the population needs that must be satisfied. In 2015, Greer and Méndez noted exactly this, as they remarked that "*there is a strong tendency to discuss universal health coverage as though it were a settled goal that only requires technical follow-up: this approach contradicts or at least underplays a large body of evidence suggesting that **universal health coverage is potentially transformative and intensely political**, and depends on the features of a country's governance*".[35] And even more recently, the October 2018 Astana Declaration adopted in the wake of the Global Conference on Primary Health Care reaffirmed "*the primary role and responsibility of governments at all levels in promoting and protecting the right of everyone to the enjoyment of the highest attainable standard of health*", and also that the path towards universal health coverage requires to "*make bold political choices for health across all sectors*".[36]

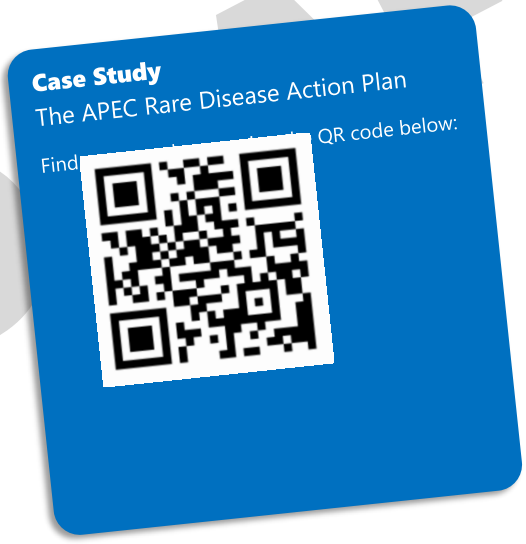
Are such "bold political choices" impossible or particularly difficult to make? Nothing could be farther from the truth, as illustrated by the recent launch of a new Rare Disease Action Plan

"No APEC economy will be able to claim that it has achieved APEC Leaders' Vision for Health, or successfully implemented 'Healthy Asia-Pacific 2020', if it has not adequately and equitably met the needs of people living with rare diseases."

Official 'Healthy Asia-Pacific 2020'

[34] https://bit.ly/2EkqtUQ
[35] https://bit.ly/2zUvrVl
[36] https://bit.ly/2ORMhNZ

by the APEC Life Sciences Innovation Forum (LSIF)'s Rare Disease Network during the APEC CEO Summit 2018 "Harnessing Inclusive Opportunities, Embracing the Digital Future", which gathered the Heads of State of the 21 Pacific Rim member economies, business leaders and innovative thinkers from 15 to 17 November 2018 in Port Moresby, Papua New Guinea. This new Rare Disease Action Plan aimed at providing all APEC Member States with a united framework for policy action to tackle the challenges of rare diseases by 2025, with a focus on 10 key areas including: raising public awareness of rare diseases; better use of technology to enhance the purposeful use of data in clinical decision-making; and healthcare system design to ensure that patients are diagnosed and cared for in a timely manner.



Note from editor: This section could be completed as of here onwards with:

- **A spotlight on the Philippines** and on the work done by PSOD (Cynthia Magdaraog) in the country to support a greater integration of rare diseases into the national healthcare system – as an illustration that, even in countries where progress towards a greater recognition of rare diseases is

more recent and emerging, there are ways to firmly embed rare diseases in a UHC perspective from the start.

- **A spotlight on South Africa** (courtesy of Kelly Du Plessis / RDSA) to highlight the emerging conversation on UHC in African countries but also the specific types of constraints that will need to be overcome in that region to make UHC a success (e.g. human resources, timeframes for actual implementation, needs-based training, balancing the external pressure to adopt and progress on UHC vs the domestic reality, etc) Focus on the practical difficulty of moving from words and principles to deeds and results on UHC, and thus, on the very importance of being even more committed to the defence of rare diseases within UHC.

Commented [A2]: Both examples to be included pending delivery of detailed content, approved by each organisation/contact person, to illustrate the proposed angles.

*

To summarise, it is our view (and, as per the preceding pages, one supported by an extensive body of literature) that:

- **Several major schools of thought in the field of social justice are strongly supportive of meeting the needs of people living with rare diseases as a moral imperative of the highest order**, especially in the context of a policy of universal scope and ambition such as universal health coverage.
- Despite recurrent concerns or doubts that fully including rare diseases in a universal health coverage approach would require dropping other no less stringent health priorities in the face of limited resources, **there is again growing counter-evidence to demonstrate that rare diseases can and must on the contrary be prioritised** if the implementation of a universal health coverage model is to stay true to its spirit.
- Finally, the over-emphasis on limited resources as a Malthusian constraint that would de facto entail the need to operate impossible choices amongst competing health priorities must be seen for what it is – an illusion and, truth be told, a "governance default" whereby political decision-makers implicitly or explicitly decline to elevate the level of priority they give to healthcare and to allocate to that particular policy sector the resources that would be needed to properly meet the needs of their population. **It is the prerogative and duty of policymakers to operate different choices, and to align their action with the overwhelming consensus that rare diseases fully belong to a universal health coverage approach.**

In the 2016 commentary article Universal Health Coverage – The Critical Importance of Global Solidarity and Good Governance[37], WHO's Andreas A. Reis observes that "*universal health coverage as a major global health goal is* ***not only a political, but also an ethical endeavour [of which] solidarity is an important underlying concept***". **Leaving rare diseases outside of current and future efforts to implement universal health coverage across the globe, or not giving them the level of priority which they deserve within these efforts, would be not just a major mistake, but a moral and intellectual betrayal of the principle of solidarity which sustains the very foundations of universal health coverage as a concept.**

The words of Helen Clark in 2016, which we quoted earlier on, now take a new resonance. But no less powerful were the words of WHO Director General Dr Tedros Adhanom Ghebreyesus in celebration of Rare Disease Day in February 2018 to reaffirm that the need to help all of the 300+ million people living with rare diseases worldwide are an essential and non-negotiable component of the spirit and of the letter of the Sustainable Development Goals and, as such, of universal health coverage too:

"The vision of the Sustainable Development Goals is a world in which no one is left behind, including people who suffer from rare diseases. Just because a disease affects a small number of people does not make it irrelevant or less important than diseases that affect millions. WHO's top priority is to support countries on the path towards universal health coverage, with the aim of ensuring that all people can access the health services they need. […] This includes access to diagnosis and treatment for people who suffer from rare diseases."

Dr Tedros Adhanom Ghebreyesus, WHO,
February 2018 [38]

[37] https://bit.ly/2CeGD0G
[38] https://bit.ly/2QXywgW

3. The Time to Include Rare Diseases is Now:

Concrete Civil Society-Driven Proposals to Advance Universal Health Coverage

DRAFT – A brief executive summary may yet be added at the start of each section.

On the path to preparing national healthcare systems for the implementation of universal health coverage, civil society organisations have a central role to play, alongside with – and in support of – national governments, international agencies, the private sector, academia and media. The International Health Partnership for UHC2030's 2018 working paper "*On the Road to UHC: Leave No One Behind – A civil society organisation perspective on how UHC can be reached by 2030*"[39] recognises that "***civil society is often best placed to gain access to, represent, and prioritise the most marginalised key populations**. **As such, civil society is a critical voice to ensure that people have the necessary access to equitable health services, that they are informed of health policies and can input into their country's health systems strengthening efforts, so that no one is excluded***".

In the same working paper like in other similar documents before it,[40] the contribution that civil society organisations can deliver to advance universal health coverage has been recognised as spanning over many dimensions – from generating local and international evidence on successful strategies to effectively influencing actual policy design and implementation in a more inclusive and participatory way, or from building more robust consensus within society on the right strategies to adopt to monitoring and evaluating their implementation by public authorities with a view to strengthening their social accountability. It has also been repeatedly acknowledged that, **without a strong involvement of civil society from the outset, the chances to achieve significant and sustainable progress on the path towards universal health coverage are curtailed**: as remarked by Ravenscroft and Marcos in The Lancet in September 2012, "*community system strengthening and community mobilisation are crucial for ensuring that universal health coverage works equally for the general population as well as for poor and marginalised groups*".[41]

Things are no different when it comes to rare diseases specifically. The global rare diseases movement which emerged decades ago has reached maturity in the course of the 2010s, is nowadays increasingly involved in the global political conversation (particularly at the UN level since the creation of Rare Diseases International in 2014 and of the NGO Committee for Rare Diseases in 2016), and finds itself now both well positioned and ready to develop and share tangible and actionable recommendations to

[39] https://bit.ly/2U8xL2G
[40] E.g. https://bit.ly/2HDDHiW
[41] https://bit.ly/2RdeY4a

catalyse the transition of current healthcare systems towards universal health coverage models that also fully take into account the needs of people living with rare diseases worldwide.

"Health systems can learn a lot from rare diseases to design systems that are fit for the future and effectively contributing to universal health coverage."

Rüdiger Krech, WHO,
11 May 2018

An illustration of this can be found in the 2018 RDI Memorandum to the WHO (April 2018, revised November 2018), intended to set ambitious goals and explore areas of concrete collaborative actions to address challenges in the field of rare diseases within the 2030 Sustainable Development Agenda and the WHO 13th General Programme of Work.

Ambition no. 1: Increase awareness of rare diseases and their visibility in healthcare systems

Ambition no. 2: Promote universal health coverage, strengthen healthcare systems and provide multi-disciplinary holistic care

Ambition no. 3: Improve access to accurate diagnosis, prevention of co-morbidities, and appropriate holistic care

Ambition no. 4: Improve access to appropriate and affordable treatment and care

Ambition no. 5: Ensure most WHO Member States implement strategies to address rare disease challenges at the national level within an international policy framework

Figure 4 · An excerpt from the 2018 RDI Memorandum to the WHO detailing its core ambitions and objectives, several of which intersect with universal health coverage

As illustrated above, whilst all of the 5 core ambitions spelled out in the 2018 RDI Memorandum to the WHO do touch upon one or more dimensions of universal health coverage to a certain extent, the second of them explicitly lays out the intention of the global rare diseases movement to "*promote universal health coverage*", and proposes to do so through a variety of strategic but very concrete approaches, e.g.:

- to improve patients' health outcomes, in terms of years of life and quality of life;

Commented [A3]: One methodological issue here as the first intent was to build heavily on the said RDI Memorandum to WHO – especially on the part of it directly related to UHC.

However, that document has not been shared in the public domain.

We could either still reference it and leave the current draft as is, whilst flagging that the document is precisely not in the public domain.

We could also remove the reference whilst still channeling much of the content of the original document into the present one ("recycling").

Alternatively, it would be fantastic if we could develop or source OTHER proposals and ideas…

- to go bottom-up, to identify and build up local capabilities, and to connect them regionally and internationally;
- to bring the existing knowledge and expertise to the patients where they are – rather than for the patients to be forced to travel abroad or far across their own country;
- to approach healthcare system organisation for rare diseases through clusters of therapeutic areas for rare diseases (around approximately 20 to 25 clusters) so as to provide an inclusive framework for all rare diseases, whilst still building step-wise upon the experience from individual rare diseases that can rely to date on more advanced clinical networks;
- to generate new knowledge through pooling of data and clinical experience wherever available.

To operationalise these approaches, RDI formulated 3 goals to be reached by the year 2030:

- **to establish a policy framework and kickstart the first pilot projects for the creation of an international network of accredited specialised centers of expertise** – a simple solution to address the fact that the most clinically relevant expertise may not always be available locally, thus creating the need for increased international cooperation and mobility of experts and patients alike, be it to establish a diagnosis, to prevent complications or to initiate treatment;
- **to leverage all digital technologies available today for enabling a greater collaboration of experts and networks** and sharing greater information amongst them, e.g. via "virtual networks" which could act as gateways to global and regional networks in a "hub-and-spoke" model, but also open up new possibilities for improved care, e.g. virtual medical consultations for persons living with a rare disease, online training and education modules for specialised healthcare professionals, or still the collection of shared common datasets and creation of patient registries;
- **to expand healthcare pathways and diagnosis, care and treatment guidelines** that exist today for well-documented rare diseases, hence fostering their faster and more uniform adoption and uptake around the globe – such "international healthcare pathways" could define the standards for safe and clinically effective diagnosis, care and treatment but also set out the minimum resources, expertise and time to optimise care and treatment – thus functioning de facto as an internationally recognised best practice or norm, which in turn would inform patient expectations and decision-making.

These proposals and goals have one merit: **they are ambitious but realistic because they have been tried and tested already. The launch of the European Reference Networks (ERN) in the European Union in early 2017 saw the formal recognition of no less than 900 expert units across the continent, connecting 370 specialist hospitals within 24 networks per major families of rare diseases** – all converging to create one single central European infrastructure firmly anchored into the national healthcare systems of 26 different Member States. These networks have helped bridge the gap between patients and experts to speed up diagnosis and drive forward the generation of new knowledge accessible by all locally. Since their launch, they have been continuing to expand in a step-wise fashion,

both in terms of partner institutions involved and in terms of numbers of rare diseases covered, ensuring that each and every individual rare disease has a home under at least one network.

European Reference Networks also provide for the first time a genuine critical mass of persons living with a rare disease, of population data and of highly-specialised clinicians and researchers, all united into one community to share expertise, knowledge and resources across borders and to drive forward research and therapeutic development.

The successful implementation of the European Reference Networks, together with the outcomes of other international initiatives and experiences in Japan, Australia or Canada still, creates a unique opportunity to adapt and roll out this template model to the next level on a global scale. Increasing the number of centers and countries connected to one another around the world, and increasing in so doing the available critical mass of patients and clinicians, will offer our best chance to serve the needs of hard-to-reach populations wherever they live, whilst making these global networks even more effective in delivering their mission.

Any other proposals from the RDI Memorandum we wish to highlight?

Provisional conclusion/endpoint for section 3:

All these proposals from civil society organisations assembled within the global rare diseases movement are only an early attempt at formulating realistic suggestions based on experience of how the needs of people living with rare diseases worldwide could be better met, in developed and developing countries alike, in an approach compatible and pursuant to the ambition of universal health coverage. **This is only the beginning, and we look forward to using these proposals as a bedrock for greater dialogue with all partners worldwide – UN agencies, national governments, other non-governmental organisations – but also as an inspiration to continue inventing new solutions that will accelerate the transformation of healthcare systems into universal health coverage ones whilst placing the needs and expectations of people living with rare diseases front and center.**

Conclusion (No working title yet…)

Reproducing for now the content of the conclusion of the Executive Summary:

> The global rare diseases movement looks forward to being represented at, and contributing to, the September 23, 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 better recognised and fully supported.
>
> We hereby call on all partners – UN agencies, national governments, other non-governmental organisations, etc – to join us in constructive conversations to explore and advance our current proposals and to ideate new ones together.

If the document in full is further developed after February and all the way up to May, we may well need to revisit that provisional conclusion in the light of developments to happen during that period!