

Submission by the



to the United Nations High Commissioner for Human Rights

for your report to the Human Rights Council at its 38th session on “*contributions of the right to health framework to the effective implementation and achievement of the health-related Sustainable Development Goals*”

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## The “Right to Health” in Rare Diseases:

### A Practical Contribution to Implementing and Achieving the Sustainable Development Goals

1 On behalf of the **NGO Committee for Rare Diseases**<sup>1</sup>, a substantive committee established under the  
2 umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO) to share  
3 knowledge about rare diseases and raise their visibility on the political stage globally, we welcome the  
4 opportunity to provide input on the relationship between rare diseases and the Sustainable Development  
5 Goals in a human rights-based framework.

6 Resolution 35/23, adopted by the Human Rights Council on 23 June 2017, enshrines “***the right of***  
7 ***everyone to the enjoyment of the highest attainable standard of physical and mental health***” not only  
8 as a central component of the 2030 Agenda for Sustainable Development, but also as an essential  
9 instrument for its actual implementation.

10 **Health intersects in a structural way several of the Sustainable Development Goals**, well beyond SDG  
11 3 only. To highlight but a few, poor health can very frequently compromise access to “quality education”  
12 (SDG 4) and to “decent work” (SDG 8), thus opening up the risk of a vicious and self-sustaining spiral  
13 leading to inequalities (SDG 10), exclusion and discrimination, particularly against women and girls (SDG 5),  
14 and ultimately poverty (SDG 1). In other words, **the highest attainable standard of physical and mental**  
15 **health represents the foundational bedrock without which an individual cannot be genuinely enabled**  
16 **to set forth on any pursuit of life**, be it towards personal, intellectual, economic or social fulfillment. Only  
17 by upholding human rights “*to health and through health*”, will the delivery of the SDGs indeed “*leave no one*  
18 *behind*”.<sup>2</sup>

19 **Such considerations are of a particularly acute relevance in the field of rare diseases**, which gather  
20 well over 6,000 conditions, often of genetic origin but also correlated to other types of infectious or  
21 environmental agents, and which affect small to ultra-small populations of patients (typically fewer than 1 in  
22 2,000 individuals). Most rare diseases are chronic, progressive, degenerative, disabling and frequently life-  
23 threatening.

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<sup>1</sup> For further information, please refer to [www.ngocommitteerarediseases.org](http://www.ngocommitteerarediseases.org). Please see [www.ngocommitteerarediseases.org/members/](http://www.ngocommitteerarediseases.org/members/) and [www.ngocommitteerarediseases.org/members/executive-board/](http://www.ngocommitteerarediseases.org/members/executive-board/) for details of the Committee’s members and of our Inception Executive Board formed 11 Nov. 2017.

<sup>2</sup> Report of the High-Level Working Group on the Health and Human Rights of Women, Children and Adolescents (2017) “*Leading the Realization of Human Rights to Health and through Health*”. [online] World Health Organisation. Available at: [apps.who.int/iris/bitstream/10665/255540/1/9789241512459-eng.pdf?ua=1](https://apps.who.int/iris/bitstream/10665/255540/1/9789241512459-eng.pdf?ua=1) [Accessed 8 Feb. 2018].

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24 Any conversation about rare diseases must be firmly grounded into their very definition, i.e. that these  
25 diseases are "rare" because their prevalence in the world population is extremely low, and because they are  
26 all too often "forgotten" precisely because of the scarcity of medical knowledge about them. **The negative**  
27 **impact of this combination of rarity and paucity of knowledge cannot be underestimated:** the lack of  
28 knowledge means that many rare diseases are not diagnosed, that disabilities and other symptoms are not  
29 recognised, and that in turn persons with rare diseases do not receive the same compensatory support (be it  
30 financial, educational or social) as those with more common and well-known conditions. **Ultimately, this**  
31 **effectively bars persons with rare diseases from genuine integration into society** and, consequently,  
32 from achieving the "*realization [...] of the economic, social and cultural rights indispensable for [their] dignity*  
33 *and the free development of [their] personality*" (art. 22 of the December 1948 Universal Declaration of  
34 Human Rights).<sup>3</sup>

35 Similarly, art. 12 of the December 1966 International Covenant on Economic, Social and Cultural Rights<sup>4</sup>  
36 enshrines "*the right of everyone to the enjoyment of the highest attainable standard of physical and mental*  
37 *health*". Most recently, the December 2006 Convention on the Rights of Persons with Disabilities<sup>5</sup> expressly  
38 recognized in its recital v "*the importance of accessibility to health in enabling persons with disabilities to fully*  
39 *enjoy all human rights and fundamental freedoms*". **However, it must also be recognized that, today,**  
40 **these noble and fundamental aspirations are far from being put in practice always and everywhere,**  
41 **and that a vast majority of persons with rare diseases remain discriminated and "left behind".**

42 Although each disease taken in isolation is rare, the challenges facing the estimated 300 million affected  
43 people worldwide and preventing their full enjoyment of the right to the highest attainable standard of  
44 physical and mental health **share strong commonalities, across diseases, across borders but also**  
45 **across the many issues at the core of the various SDGs**, as detailed in figure 1:

|  |   |  |   |
|--|---|--|---|
|   | Persons with rare diseases are often trapped in a vicious cycle of <b>vulnerability</b> and <b>poverty</b> due to <b>exclusion</b> from healthcare and education systems, as well as the job market.  |                   | Persons with rare diseases and disabilities are often <b>marginalised from the job market</b> not only due to prejudice, stigma and discrimination, but also due to <b>lack of accessible facilities</b> , of <b>flexible working hours</b> and of adapted roles <sup>6</sup> . |
|   | Achieving <b>universal health coverage</b> requires catering to the needs of persons with rare diseases, who need more and better medicines, appropriate diagnosis and lifelong care and social support, as well as access to assistive technologies. |                   | With no more than a few hundred developed therapies vs. nearly 7,000 repertoried diseases, <b>the need remains immense to invest in R&amp;D</b> , health technologies and diagnostic tools for persons with rare diseases.  |
| Achieving good health for persons with rare diseases is also often made difficult by <b>the lack of awareness of these diseases amongst healthcare professionals</b> , which leads to <b>delays in diagnosis</b> (or even misdiagnosis), <b>discrimination and stigma</b> . <sup>7</sup> <b>The relative invisibility of rare diseases in healthcare systems</b> (exemplified by their under-representation in the WHO International Classification of Diseases <sup>8</sup> ) also poses a major barrier. |   | <b>Accessibility to infrastructure</b> also must be improved to enable genuine inclusion in society. |   |

<sup>3</sup> See [www.un.org/en/universal-declaration-human-rights/](http://www.un.org/en/universal-declaration-human-rights/)

<sup>4</sup> See [www.ohchr.org/EN/ProfessionalInterest/Pages/CESCR.aspx](http://www.ohchr.org/EN/ProfessionalInterest/Pages/CESCR.aspx)

<sup>5</sup> See [www.un.org/disabilities/documents/convention/convoptprot-e.pdf](http://www.un.org/disabilities/documents/convention/convoptprot-e.pdf)

<sup>6</sup> 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 Anierte, 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](http://www.ilo.org/madrid/fow/trabajo-decente-para-todos/WCMS_548585/lang-es/index.htm) [Accessed 8 Feb 2018].

<sup>7</sup> See "*The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe*" [online] EURORDIS-Rare Diseases Europe. Available at: [www.eurordis.org/publication/voice-12000-patients](http://www.eurordis.org/publication/voice-12000-patients) [Accessed 8 Feb 2018].

<sup>8</sup> "*Does Your Rare Disease Have a Code?*" [online] EURORDIS-Rare Diseases Europe. Available at: [www.eurordis.org/news/does-your-rare-disease-have-code](http://www.eurordis.org/news/does-your-rare-disease-have-code) [Accessed 8 Feb. 2018].

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*Figure 1 – A brief overview, across SDGs, of the multifaceted challenges and forms of discrimination faced by persons with rare diseases preventing their enjoyment of the right to the highest attainable standard of physical and mental health*

46 **Several of the issues and challenges faced by persons with rare diseases also intersect major**  
47 **dimensions of the “right to health” framework, not least of which:**

- 48 • **Accessibility** is very often one of the most severe limitations faced by persons with rare diseases,  
49 and one impacting all facets of an individual's life. Accessibility bottlenecks are often experienced in  
50 the field of:
- 51 – proper and timely **access to treatments and health services and care**<sup>11</sup>
  - 52 – **access to understandable information** about an individual's own condition and to an  
53 accurate and timely diagnosis<sup>12</sup>
  - 54 – **access to infrastructure**, and even more broadly of access to equal opportunities (e.g.  
55 education, career, reproductive health, etc).

<sup>9</sup> 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](http://portal.unesco.org/en/ev.php-URL_ID=31058&URL_DO=DO_TOPIC&URL_SECTION=201.html) [Accessed 8 Feb. 2018].

<sup>10</sup> 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](http://www.eurordis.org/news/3000-rare-disease-patients-carers-voice-difficulties-balancing-care-life) [Accessed 8 Feb. 2018].

<sup>11</sup> See “Access to treatment: Unequal care for European rare disease patients” [online] Available at: [download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf](http://download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf) [Accessed 8 Feb. 2018].

<sup>12</sup> See “Estudio sobre situación de Necesidades Sociosanitarias de las personas con Enfermedades Raras en España – Estudio ENSERio” [online] Available at: [www.enfermedades-raras.org/images/stories/documentos/Estudio\\_ENSERio.pdf](http://www.enfermedades-raras.org/images/stories/documentos/Estudio_ENSERio.pdf) [Accessed 8 Feb. 2018].

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- Similarly, *availability* remains very much an aspiration and a pressing need for the global rare disease community, in a time when new treatments for an immense majority of the rare diseases known to this day are yet to be invented, when more must be done to facilitate the procurement and distribution of those medicines to those population groups and in those countries where they are needed most, or when public information about rare diseases could also be made more widely available to the general public. The reality experienced by persons with rare diseases is often still very far from ideal – **recent findings indicate that nearly a quarter of persons with rare diseases in Europe did not get the medical treatment they needed in the past year because the treatment was not available in their country.**<sup>13</sup>
  - The objective of *participation* is one around which all of the global rare disease community is fully mobilised today, yet it is also an objective gravely compromised and made unattainable by the frequent marginalisation of persons with rare diseases as an “invisible” population, by the burden of caregiving duties often borne by their relatives, and by the overall discrimination and stigma still often associated with rare diseases in our societies.
  - Persons with rare diseases share **the same inalienable right as all others** to life, health, *good quality* of life and holistic development and *good quality* of treatment and care.
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72 These aspects have been amply **recognised by the UN Committee on Economic, Social and Cultural**  
73 **Rights (CESCR)** in its General Comment No. 14 on “*The Right to the Highest Attainable Standard of*  
74 *Health*”<sup>14</sup> from which it can be directly derived that the “right to health” means that persons with rare diseases  
75 must have a sufficient availability of promotive, preventive, curative, rehabilitative and supportive/palliative  
76 healthcare services, medicines, health devices and technologies that are of a reasonable standard of quality,  
77 safety, accessibility, acceptability and affordability, and that are delivered to them without discrimination or  
78 financial ruin.

79 We also contend that, from a broader perspective, **the**  
80 **specific nature of rare diseases also calls for a holistic,**  
81 **comprehensive and multi-disciplinary response, deeply**  
82 **grounded by essence in a human rights vision.** This  
83 very need is starting to receive wider recognition and  
84 support, as illustrated by the reflections of UN Special  
85 Rapporteur on the Right to Health Prof. Dainius Puras on  
86 the occasion of the Rare Diseases International Policy  
87 Event “*The Right to Health: The Rare Disease Perspective*”  
88 on 10 February 2017 in Geneva.<sup>15</sup>

***"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, Administrator, United Nations  
Development Programme (Oct 2016)<sup>16</sup>

89 **Rare diseases must be addressed as a human rights and public health issue insofar as:**

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- Persons, and particularly children, affected by rare diseases have a right to life, health, good quality of life and holistic development and good quality of treatment and care
  - Existing gaps in research, early identification and treatment of rare diseases need to be closed
  - More must be done to encourage the research and development of needed medicines, and to facilitate the procurement and distribution of those medicines to those population groups and in those countries where they are needed most

<sup>13</sup> See “Access to treatment: Unequal care for European rare disease patients” [online] Available at: [download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf](https://download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf) [Accessed 8 Feb. 2018].

<sup>14</sup> As adopted at the 22nd CESCR Session on 11 August 2000, and accessible here: [www.refworld.org/pdfid/4538838d0.pdf](http://www.refworld.org/pdfid/4538838d0.pdf)

<sup>15</sup> See “*The Right to Health and Rare Diseases*” [online] Available at: [download.eurordis.org.s3.amazonaws.com/rdi/2.Puras%20Presentation-%20Rare%20diseases.pptx](https://download.eurordis.org.s3.amazonaws.com/rdi/2.Puras%20Presentation-%20Rare%20diseases.pptx) [Accessed 8 Feb. 2018].

<sup>16</sup> As per her written statement at the 11th Annual International Conference on Rare Diseases and Orphan Drugs, accessible here: [www.undp.org/content/undp/en/home/presscenter/speeches/2016/10/20/helen-clark-written-statement-at-the-11th-annual-international-conference-on-rare-diseases-and-orphan-drugs.html](http://www.undp.org/content/undp/en/home/presscenter/speeches/2016/10/20/helen-clark-written-statement-at-the-11th-annual-international-conference-on-rare-diseases-and-orphan-drugs.html)

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- 96 • States and public authorities must ensure existing medicines, other non-pharmacological treatments  
97 and assistive devices are affordable and accessible on a non-discriminatory basis, and where  
98 necessary in low-cost forms or versions for low-resource countries<sup>17</sup>
- 99 • A comprehensive approach to rare diseases is needed, encompassing education, research,  
100 prevention, diagnosis, care and treatment, social support and inclusion
- 101 • Services and support for patients and families affected by rare diseases need to be holistic and  
102 integrated to provide for the health, disability and social issues often associated with them
- 103 • Specific funding from governments for research projects on rare diseases is needed
- 104 • International patient registries and biospecimen repositories for rare diseases should be encouraged  
105 and available for appropriate use by the rare diseases community

### 106 **To conclude, and based on the preceding elements, it is our firm belief that:**

- 107 • There is a pressing need for the specificities of persons with rare diseases to be better  
108 understood and recognised, at all levels, so that such minority groups cease to be subject to  
109 discrimination, neglect or exclusion. This is an absolute necessity if the principle that underpins  
110 the entire 2030 Agenda for Sustainable Development – “*to leave no one behind*” – is to be  
111 successfully advanced from aspiration to reality, to the benefit of all mankind.
- 112 • **States have a crucial duty to raise awareness and increase knowledge about rare**  
113 **diseases**, and about the profound everyday consequences faced by affected individuals. This  
114 cannot be underestimated as professionals in the areas of healthcare, social services,  
115 education and employment – on whom persons with rare diseases depend in every single  
116 aspect of their everyday lives – very often do not have sufficient information, knowledge or  
117 specialised competencies to properly handle and meet the full range of implications and special  
118 needs that rare diseases do create.
- 119 • **Several of the underlying principles of the “right to health” framework bear a close**  
120 **correspondence to well-identified bottlenecks and challenges which must be addressed**  
121 **in order to progress towards several important SDGs** and to deliver the full magnitude of the  
122 ambitions of the 2030 Agenda for Sustainable Development.
- 123 • **Rare diseases offer a unique and privileged terrain for the practical application of the**  
124 **“right to health” framework** towards the realisation of the objectives assigned to the SDGs,  
125 but also for the definition of concrete solutions that will offer tangible metrics of progress on the  
126 path to 2030.
- 127 • **Even more fundamentally, the right approach to address the global challenge of rare**  
128 **diseases today has to be a human rights-based one**, which only increases the relevance of  
129 rare diseases in the current and future implementation of the “right to health” framework and of  
130 the SDGs.
- 131 • While the vast majority of existing initiatives and efforts to deliver the objectives of the SDGs  
132 may benefit persons with rare diseases to a certain extent, in many cases they may not  
133 because of the unique nature of these small populations whose conditions are often  
134 undiagnosed, whose symptoms are not acknowledged, and whose disease progression is  
135 undocumented. **Specific compensatory actions are required, both in the form of strategies**  
136 **and frameworks addressing all rare diseases, and/or in the form of distinct actions for**  
137 **rare diseases under and within each SDG.**

<sup>17</sup> Further to the objectives of the World Health Organisation's Global Cooperation on Assistive Technology (GATE) Project, information on which can be accessed here: [www.who.int/phi/implementation/assistive\\_technology/phi\\_gate/en/](http://www.who.int/phi/implementation/assistive_technology/phi_gate/en/)