



NGO COMMITTEE FOR
RARE DISEASES

REPORT FROM
RARE DISEASE DAY®
POLICY EVENT AT THE
UNITED NATIONS

SECOND HIGH LEVEL EVENT OF THE NGO
COMMITTEE FOR RARE DISEASES



Organised by

ÅGRENKA



EURORDIS
RARE DISEASES EUROPE



RARE
DISEASES
INTERNATIONAL

An event to mark the occasion of



Under the patronage of

**HRH The Grand Duchess
of Luxembourg**



© 2018 Cour
grand-ducale / Lola Velasco

Co-Hosted by



KINGDOM OF BELGIUM
www.diplomatie.belgium.be



**PERMANENT MISSION OF
THE REPUBLIC OF CYPRUS
TO THE UNITED NATIONS**



**PERMANENT MISSION
OF ESTONIA TO THE UN**



**Mission permanente de la France
auprès des Nations unies**



**PERMANENT MISSION
OF JAPAN TO THE
UNITED NATIONS**



**Permanent Mission of the
State of Kuwait to the United Nations
New York**



**GRAND DUCHY OF LUXEMBOURG
Permanent Mission to
the United Nations**



**PERMANENT MISSION OF
THE REPUBLIC OF MALTA
TO THE UNITED NATIONS**



**PERMANENT MISSION OF ROMANIA
TO THE UNITED NATIONS**



**PERMANENT MISSION
OF THE REPUBLIC
OF SERBIA TO THE
UNITED NATIONS**



**MISIÓN PERMANENTE
DE ESPAÑA ANTE
LAS NACIONES UNIDAS**



**Permanent Mission of Sweden
to the United Nations, New York**



**PERMANENT MISSION
OF THAILAND TO THE
UNITED NATIONS**



**PERMANENT MISSION OF
THE UNITED ARAB EMIRATES
TO THE UNITED NATIONS
NEW YORK**

With the support of



Co-funded by
the Health Programme
of the European Union



The NGO Committee for Rare Diseases: working on a common cause at the global level

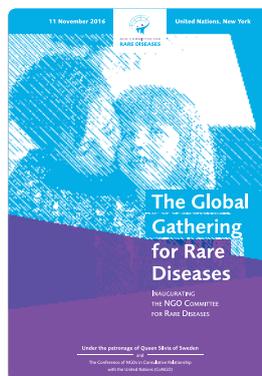
The NGO Committee for Rare Diseases is a substantive committee established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO). It was initiated by the Ågrenska Foundation and EURORDIS-Rare Diseases Europe, with the later integration of Rare Diseases International (RDI), and its formation was approved by a vote of 27 CoNGO member organisations in April 2014. Its goal is to promote rare diseases as a priority in global health, research, and social and medical care as part of the UN 2030 Agenda: the Sustainable Development Goals (SDGs).

Why the United Nations? The Committee aims to work with the United Nations as this intergovernmental organisation has been, over the decades, a driver for a strong public health agenda, social development, inclusion of vulnerable populations, development in science, technology and innovation, and human rights for all. As such, it is a key enabler for better lives for persons living with a rare disease.

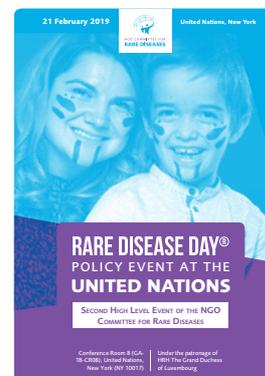
The NGO Committee for Rare Diseases held its first high-level event and official inauguration, 'The Global Gathering for Rare Diseases', in November 2016 at the United Nations Headquarters in New York, under the patronage of Queen Silvia of Sweden and with the support of five Member States. On that occasion, the link between the rare disease community's goals and the UN SDGs was established and recognised by the main UN agencies and bodies, which opened the door to broader awareness of the issue and potential partnership at global level.

The second high-level event of the NGO Committee for Rare Disease, 'Rare Disease Day Policy Event at the United Nations', took place on 21st February 2019, to mark the occasion of Rare Disease Day. It was held under the patronage of The Grand Duchess of Luxembourg, hosted by the Permanent Mission of Estonia to the United Nations and co-hosted by 14 Permanent Missions from: Belgium, Brazil, Cyprus, France, Japan, Kuwait, Luxembourg, Malta, Romania, Serbia, Spain, Sweden, Thailand, and the United Arab Emirates. The meeting took the form of an all-day roundtable bringing together around 100 participants –

from the international NGO community, UN agencies, national governments, academic and research institutions as well as the private sector – interested in collaborating towards the advancement of rare diseases as a global public health priority within the United Nations.



NGO Committee for Rare Diseases first high-level event (11th November 2016)



NGO Committee for Rare Diseases second high-level event (21st February 2019)



From left to right: Hatijah Ayob (Malaysian Rare Disorders Society), Irina Miasnikova (Russian Association for Rare Diseases) and Eda Selebatso (Botswana Organisation for Rare Diseases)



Rare Disease Day Policy Event taking place in Conference Room 8, United Nations Headquarters in New York

Recognising rare diseases as a policy priority: empowering people living with a rare disease

There are over 6,000 identified rare diseases. Even though one disease may be rare, the number of people affected by rare diseases is extremely large, with an estimated over 300 million worldwide.

Rare diseases are often chronic, highly complex, progressive and severely disabling, frequently affecting life expectancy and generating specific care needs. As such, people living with a rare disease face various challenges in accessing treatments, healthcare and social care. Indeed, the patient population for each disease is small and the expertise is usually limited. Many people remain undiagnosed, misdiagnosed or wait years for a diagnosis.

the healthcare, research, social care, school and working environments, but also with policy makers and with society at large.

An essential step, as Dr. Ana Rath, Director of Orphanet, mentioned, is that awareness within the healthcare field is translated into visibility in health information systems in which rare diseases are currently not well represented. Quoting Dr. Gareth Baynam, Dr. Rath said: "if we cannot count rare disease patients, rare disease patients do not count". In particular, she explained that health information systems need to integrate a rare disease-specific codification, since the latter enables measuring the

A rare disease affects...



HEALTH



FAMILY



EDUCATION



WORK



ECONOMY



SOCIETY



SCIENCE

Furthermore, rare diseases impact not only a person's health, but also their socio-economic status, family, education and labour opportunities. At the event, Lieven Bauwens, Secretary General of the International Federation for Spina Bifida and Hydrocephalus, presented the results of a recent Rare Barometer Voices survey on the effects of rare diseases on daily life proving that difficulties such as poverty, unemployment and social exclusion are in fact a daily reality for most people affected. People with a rare disease thus fall under the same category as traditionally disadvantaged groups, being a vulnerable and marginalised population, with the added 'specificity' of the rarity.

All the struggles mentioned above are shared across many different diseases and geographies, making rare diseases a global health priority demanding common solutions. This entails on the one hand, the need to consider rare diseases as a collective grouping rather than looking solely at each individual disease, and on the other hand, the need to collaborate across borders and worldwide. No one country, no one continent alone can address alone the issues created by rare diseases.

In this context, as Avril Daly, Chief Executive Officer of Retina International and Vice-President of EURORDIS-Rare Diseases Europe, described at the event, giving visibility to people living with a rare disease is the first step to take in order to enable inclusion in society and the development of policies. Awareness must be raised with professionals in

burden of rare diseases on the population, focusing health and social service delivery on specific patients' needs, and identifying patients for clinical research. Orphanet is in fact producing this specific nomenclature with unique identifiers called 'orphaCodes', and this is already being implemented successfully in various countries. In addition, Orphanet is a WHO Collaborative centre working to integrate rare



Not being visible is the first obstacle to recognition and to access good standards of care, but also for policy makers, it hampers building appropriate strategies based on evidence. »

Ana Rath, Director, Orphanet



diseases in ICD-11 (International Classification of Diseases 11th Revision). Dr. Rath also presented an upcoming study by Orphanet that will give clarity on the figures of persons living with a rare disease worldwide, thus facilitating the appraisal of rare diseases as a public health issue. For instance, she explained that results seem to point out that the large majority of persons affected (>98%) has in fact one of the 390 most prevalent rare diseases. This means that setting up strategies for rare diseases as a collective grouping is sound public health policy and in fact can

is essential, and both enrich the projects and accelerate the pace of research in ways that would otherwise be impossible at local level.

Collective mobilisation for stronger visibility, recognition and international collaboration in support of rare diseases has to date translated into practice in various fields. For instance, in medical research, initiatives like the International Rare Diseases Research Consortium (IRDiRC) have emerged. At the industry level, a dedicated Working



Sharing data is one of the most important areas when it comes to rare diseases, because with so few patients, it is very difficult to do anything for one institution, for one country or one region »

Dr. Irene Norstedt, Acting Director responsible for the Health Directorate within the Directorate-General for Research and Innovation, European Commission and Head of Unit, Innovative and Personalised Medicine Unit; Member of the International Rare Diseases Research Consortium (IRDiRC)

allow for the creation of frameworks of expertise that serve patients with the rarest diseases as well.

In the field of research, Dr. Irene Norstedt of the European Commission Directorate-General for Research and Innovation, highlighted how cross-fertilisation and collaboration with different stakeholders at global level

Group on Rare Diseases has been created within the International Federation of Pharmaceutical Manufacturers & Associations (IFPMA). This is all facilitated by the creation of a strong global rare disease patient community through initiatives like Rare Diseases International (RDI), RareConnect and Rare Disease Day.



The second high-level event of the NGO Committee for Rare Diseases took place within the Rare Disease Day campaign. Rare Disease Day was coined on February 29th 2008 – a 'rare' date – however the campaign is now celebrated every year on the last day of February. Rare Disease Day acts as an enabler and its main objective is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on people's lives.

Since Rare Disease Day was first launched by EURORDIS in 2008, thousands of events have taken place throughout

the world, reaching hundreds of thousands of people and resulting in a great deal of media coverage. The first Rare Disease Day was celebrated in 18 countries (all European). In 2019, for its 12th edition, events took place in over 90 countries and regions from around the world.

Rare Disease Day has helped in creating a sense of community across the different rare diseases, generating a common identity as well as an international momentum. At the event, Terkel Andersen, President of EURORDIS-Rare Diseases Europe, issued a plea on behalf of the rare disease community for Rare Disease Day to become an official awareness day of the United Nations. In this way, the last day of February would become a unique opportunity for even stronger communication and advocacy campaigns, giving people living with a rare disease the global recognition they deserve.



Yukiko Nishimura and Kunihiro Nishimura (Advocacy Service for Rare and Intractable Diseases' multi-stakeholders in Japan (NPO ASrid)), Terkel Andersen (EURORDIS-Rare Diseases Europe) and Dora Maradiaga (NCD Alliance)

Rare Diseases within the United Nations Sustainable Development Goals (SDGs)



We must keep in mind that our main goal should be that of Agenda 2030 - that is the imperative of leaving no one behind, which, by definition, also includes leaving no person living with a rare disease behind. 2030 Agenda is for all people, and our work here is to ensure that we will live up to this precious promise »

H.E. Sven Jürgenson, Ambassador and Permanent Representative of Estonia to the United Nations

Rare Disease Day Policy Event 2019 solidified the clear alignment between rare diseases and a number of Sustainable Development Goals (SDGs), as well as the 2030 Agenda's guiding principle 'to leave no one behind'.

The 2030 Agenda for Sustainable Development was adopted by the UN General Assembly in September 2015. It includes 17 goals, the SDGs, each of which consists of targets that should be achieved by countries within the next 15 years. SDGs address global challenges such as poverty, health, climate, peace and justice, and should be viewed in a holistic way. They are interdependent and are to be achieved by both developing and developed countries. In this sense, they are universal.

During the meeting, the relevance of rare diseases within the SDGs was reiterated. In particular, the fact that this relevance goes beyond just SDG3 on health and touches also upon SDGs on education (SDG 4), gender (SDG 5), work (SDG 8), inequality (SDG 10) and partnerships (SDG 17) was stressed.

Robert Hejdenberg, Chief Executive Officer of Ågrenska Foundation, clearly explained this concept: 'the symptoms of a rare disease lead to complex consequences that go far beyond the strictly medical situation, affecting many different areas of a person or a family's life'. As a consequence, 'the individual requires support by different parts of society: healthcare, labour, social care, school, insurance'. For this reason, to ensure the well-being of people living with a rare disease, it is necessary to think beyond the health system and to promote measures that are multidisciplinary, holistic and person-centred, and that ensure non-discrimination and opportunities to contribute to society.

A particularly important outcome of the event was the explicit support of main bodies and agencies of the United

Nations (OHCHR, ECOSOC & DESA, WHO) as well as Member States towards this notion. This is tangible proof of the progress made since the launch of the NGO Committee for Rare Diseases in 2016 when the idea of securing the place of rare diseases within the UN 2030 Agenda was first introduced. Leslie Wade, from the UN Department of Economic and Social Affairs, commended the NGO Committee for taking the full measure of what the SDGs can mean for people with a rare disease and, how addressing their plight will contribute to realise the SDGs.

In the last two years, a series of advocacy actions have been piloted through the NGO Committee for Rare Diseases within the human rights, health, and sustainable development agendas of the UN system, further reinforcing this key message and bringing together allies and 'champions' towards this cause. These include:

HUMAN RIGHT TO HEALTH

- » Submission of '**The Right to Health in Rare Diseases**' to the open consultation by the Office of the High Commissioner for Human Rights (OHCHR) for the 'Report on the contributions of the right to health framework to the effective implementation and achievement of the health-related SDGs' (June-July 2018).
- » Official Written Statement on '**The Human Rights of People living with a Rare Disease**' to the 38th Session of the Human Rights Council (June-July 2018).

THE RIGHTS OF PERSONS WITH DISABILITIES:

- » Participation in the '**Expert Group Meeting on the right of persons with disabilities to the highest attainable standard of physical and mental health**' at the OHCHR, Geneva (May 2018).

- » Oral statement delivered at the '**Conference of State Parties to the Convention on the Rights of Persons with Disabilities**' in New York (June 2018).
- » Contribution to the '**Report of Special Rapporteur on the Right of People with Disabilities on the right to the highest attainable standard of health**' (OHCHR, October 2018). In the Report it is said that a 'lack of accurate diagnosis and effective treatments for the majority of rare diseases represents a challenge worldwide'. Moreover, the Special Rapporteur called on States to 'consider developing and implementing policies and practices targeting the most marginalized groups of persons with disabilities', including rare diseases.

NON COMMUNICABLE DISEASES (NCDs):

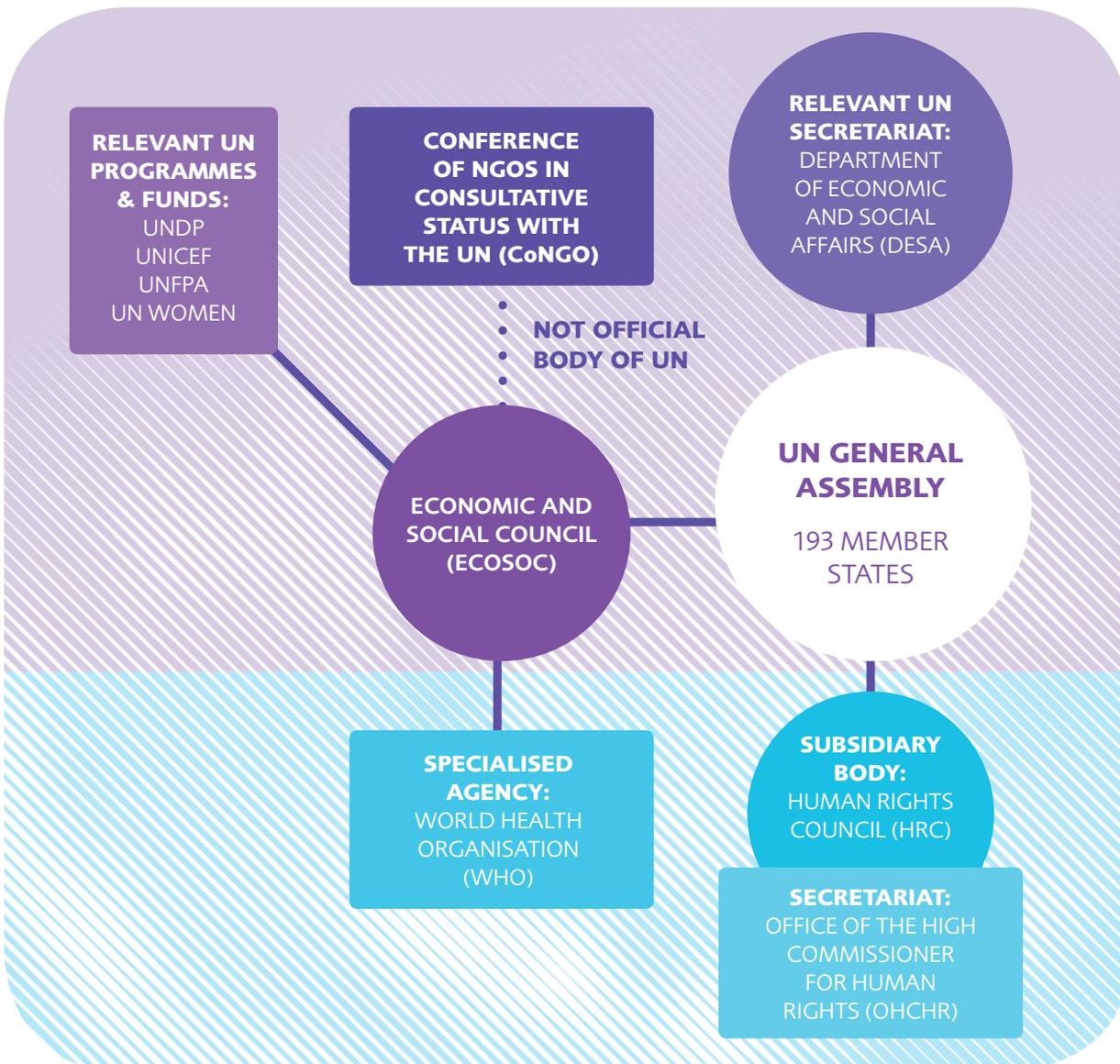
- » Submission to the **consultation of the WHO Independent High-Level Commission on NCDs** (May 2018).
- » Participation of RDI to the UN NCD Civil Society Hearing in New York (July 2018).

UNIVERSAL HEALTH COVERAGE (UHC)

- » Participation in the **High Level Panel organised by WHO and the UN Group of Friends of UHC** to celebrate Universal Health Coverage Day on 12th December 2018 in New York.
- » Preparation towards the **High Level Meeting on UHC** at the UNGA in September 2019, including the participation in the Multi-Stakeholder Hearing on UHC, on April 29th 2019.

In parallel, the NGO Committee has supported the advocacy messages of Rare Diseases International (RDI) towards the World Health Organisation (WHO), which included an official statement delivered at the **71st World Health Assembly** in Geneva (May 2018) and a response to the open consultations on **WHO's 13th General Programme of Work 2019-2023**. Closer collaboration between RDI and WHO is on-going and foreseen for the years to come and the NGO Committee will continue to support this endeavour and disseminate those messages to other UN actors.

Simplified diagram of UN actors relevant to the work of the NGO Committee for Rare Diseases





Alain Weill, President of World Hemophilia Federation and member of board of NGO Committee for Rare Diseases, and Dr. Ruediger Krech, Director, Universal Health Coverage and Health Systems, World Health Organisation



The lack of adequate investment in research and innovation, access to diagnostics, medicines and treatments for the 6000 conditions that are considered rare is a clear threat to the principle of leaving no one behind »

Leslie Wade, Chief, Inter-organizational and Inter-Institutional Support Branch, UN Department of Economic and Social Affairs



SDG 3 is about ensuring that people can live long and healthy lives. It is about making sure that even with a disease you have the support you need to be healthy. You can live with a disease and still be healthy, be productive and contribute to the economic development of the country »

Anders Nordström, Ambassador Global Health, Swedish Ministry of Foreign Affairs



The synergies between the UN Sustainable Development Goals and rare diseases

SDGS	RARE DISEASE CHALLENGES	SDGS	RARE DISEASE CHALLENGES
<p>1 NO POVERTY</p>	Persons with a rare disease are often trapped in a vicious cycle of vulnerability and poverty due to exclusion from health care and education systems, as well as job markets	<p>8 DECENT WORK AND ECONOMIC GROWTH</p>	Persons with a rare disease and disabilities are often marginalised from job markets due to lack of accessible facilities, flexible working hours and adapted roles
<p>3 GOOD HEALTH AND WELL-BEING</p>	Persons with a rare disease need more and better medicines, appropriate diagnosis and lifelong care and social support	<p>9 INDUSTRY, INNOVATION AND INFRASTRUCTURE</p>	<p>There is a need to invest in research & development of therapies, health technologies and diagnostic tools</p> <p>Accessibility to infrastructure is key to be included in society</p>
<p>4 QUALITY EDUCATION</p>	50% of rare diseases affect children who often face difficulties attending school due to inaccessibility of facilities and non-adapted teaching methods	<p>10 REDUCED INEQUALITIES</p>	Persons with a rare disease tend to remain a marginalised population suffering from discrimination in the health, labour and governance fields
<p>5 GENDER EQUALITY</p>	Gender equality means recognising and valuing unpaid care and domestic work that many mothers of children with rare diseases (including when they are grown adults) take on	<p>17 PARTNERSHIPS FOR THE GOALS</p>	The rare disease community is increasingly interconnected, with a myriad of networks of patient advocates, regulators, research & industry

Commitment towards the integration of Rare Diseases within Universal Health Coverage (UHC)

According to the WHO, Universal Health Coverage (UHC) means 'that all individuals and communities receive the health services they need without suffering financial hardship'. In other words, UHC enables everyone to access the services that address the most significant 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. The concept includes the full spectrum

The essence of the aspirations and objectives behind UHC strongly resonate with the daily experience, needs and expectations of people living with a rare disease across the world. The rare disease community and its representatives have long advocated for the right of individuals to receive the right health services without hardship and have emphasised the need to move towards a holistic and integrated approach to health.



The exclusion of rare diseases and conditions from medical coverage schemes is a violation of the right to health. As Member States move to seek consensus on a Political Declaration on UHC later this year, we need to ensure that rare diseases are covered and that people living with them benefit from this protection »

Andrew Gilmour, Assistant Secretary-General for Human Rights and Head of the Office of the High Commissioner for Human Rights in New York

of essential, quality health services, from health promotion to prevention, treatment, rehabilitation, and palliative care. All these feature make of UHC a catalyst for socio-economic development and a key contributor to equity, social justice and inclusive growth economy.

All UN Member States have agreed to work towards achieving UHC by 2030, as part of SDGs. In particular, the General Assembly Resolution on 'Transforming our world: the 2030 Agenda for Sustainable Development' adopted in 2015 emphasises that UHC is a core target of SDG 3 on health. This entails that unless major efforts are put in place to achieve UHC by 2030, the international community will fail to deliver on its promise to achieve SDG 3.



Dr. Satoshi Ezoe, Counsellor on Health at the Permanent Mission of Japan to the UN presents the Nan-Byo Strategy in Japan, a country where measures for rare diseases have been taken since the 1970s, with an official Act enacted in 2014

In spite of this, rare diseases have been largely left out from the political conversations on UHC until recently. However, UHC shall never be fully realised if persons affected by a rare disease are left behind and their needs are not met.

The Rare Disease Day Policy Event served as a platform to show that rare diseases integrally belong, by their very



UHC embodies the notion of 'No One Left Behind', the core principle of the SDGs, and underpins the concept of human security, which Japan fundamentally subscribes to. In that regard, we will not have achieved Universal Health Coverage unless and until we ensure that persons with rare diseases are not left behind »



H.E. Toshiya Hoshino, Ambassador and Deputy Permanent Representative of Japan to the United Nations



Dr. Simon Kos, Chief Medical Officer and Senior Director, Microsoft & member of the Global Commission on ending the Diagnostic Odyssey of Children with Rare Diseases, presenting new opportunities to improve the diagnosis of children living with a rare disease

nature, to the concept of UHC. In fact, discussions at the event focused on the need to integrate rare diseases into the upcoming landmark UN Political Declaration on UHC, due to be adopted at the first ever High-Level Meeting on UHC during the UN General Assembly in September 2019. To this end, the NGO Committee for Rare Diseases International is supporting a new position paper from Rare Diseases International that will serve to push towards this objective.

Ambassadors and UN officials at the event gave encouraging statements towards this goal, including Assistant Secretary-General for Human Rights Andrew Gilmour, and the Ambassador of Thailand H.E. Vitavas Srivihok, who is the co-facilitator of consultations on the Political Declaration on UHC with Member States.

Furthermore, during the afternoon plenary, national strategies, case-studies and pilots put in place in a number of Member States and carried in collaboration with organisations of people living with a rare disease and/or with governments support were showcased. Countries from across regions of the world presented their strategies, including Japan, France, Colombia, Canada, United States, Romania, Iran, China, Brazil, Philippines, South Africa and Kuwait as well as the economies within APEC (Asia-Pacific Economic Cooperation). The meeting made clear that, even in resource-constrained settings, the implementation

Cynthia Magdaraog, President of the Philippine Society for Orphan Disorders, Inc. presents the consolidated UHC Bill of 2019 which integrates measures to address the needs of persons living with a rare disease in The Philippines



« We must work together to ensure an equitable and affordable access to quality health services and medicines for everyone, especially people who are most in need, which also includes people living with rare diseases, among others, while ensuring that they do not face financial hardship or fall back into poverty because of their catastrophic medical expenses »

H.E. Vitavas Srivihok, Ambassador and Permanent Representative of Thailand to the United Nations (Co-facilitator of consultations with Member States ahead of UHC High-Level Meeting)

of national policies on rare diseases not only is possible, but also definitely contribute towards UHC and towards inclusive and equitable societies.

These developments across the world offer hope and reassurance that national governments and regional organisations are gradually realising the importance of rare diseases in a Universal Health Coverage context, and adopting new policies and legislations in this regards.



« We need to secure that as we move to the GA HLM in September, we are making sure that rare diseases are finding their deserved place in these conversations and in the commitments that each head of State will hopefully make. WHO stands by you with this commitment and we will be as always making sure that this issue is not forgotten »

Dr. Nata Menabde, World Health Organisation NYC Office Director

Call for a United Nations General Assembly Resolution on Rare Diseases

The Second High-Level Meeting of the NGO Committee for Rare Diseases launched a call for the UN to adopt a resolution at the General Assembly (GA) that will formally elevate the cause of rare diseases and promote global collaboration to overcome the barriers of rarity. By doing so, it will set in motion a wave of policy actions that will ultimately help address the challenges in an holistic approach to improve the lives of persons affected by rare diseases around the world.

UN General Assembly Resolutions are voted on by all Member States of the UN in the General Assembly, and usually require a simple majority to pass. They reflect the views of the Member States, provide policy recommendations, assign mandates to the UN Secretariat and the subsidiary bodies of the General Assembly and decide on all questions regarding the UN budget. They are generally not-binding for Member States, and the implementation of the policy recommendations contained in them is the responsibility of each Member State. Yet, the General Assembly remains the preeminent space for international dialogue and discussion, and its resolutions express the views of the international community on a certain issue. In addition, GA resolutions carry a strong political message and can significantly influence Member States' behaviour. In fact, Member States themselves often use GA resolutions as instruments of persuasion and de facto soft law.

For these reasons, a UN GA Resolution on Addressing Holistically the Challenges of People Living with a Rare Disease would be a remarkable step ahead towards global recognition and actions towards the improvement of their overall life conditions. Indeed, the Resolution would become a point of reference to substantiate the claims of the rare disease community in front of Member States and could help the development of national strategies as well



Our vision is that in order to meet the needs of the rare disease community, it is absolutely necessary to develop, adopt and implement national plans in this field »

H.E. Ion Jinga, Ambassador and Permanent Representative of Romania to the United Nations (on behalf of the Romanian EU Presidency)



as the creation of international cross-border collaboration. All in all, a Resolution on Rare Diseases would therefore also contribute to the achievement of the SDGs and the ambition to leave no one behind.

During the event, Yann Le Cam, Chief Executive Officer of EURORDIS-Rare Diseases Europe and member of the Council of Rare Diseases International (RDI) and of the Board of the NGO Committee for Rare Diseases, issued the call for a GA Resolution on Rare Diseases on behalf of these bodies, 'which are the expression of civil society on rare diseases within ECOSOC and the UN system, acting as proxies of the 300 million living with a rare disease'.

Martin Seychell, Deputy Director-General for Health and Food Safety at the European Commission, presented the European Reference Networks (ERNs), a groundbreaking model to overcome scarcity and scattering of expertise for rare diseases





From left to right: Prof. Matthew Bellgard, Director of eResearch, Division of Research and Innovation, Queensland University of Technology & Chair of APEC LSIF Rare Disease Network; Germán Escobar Morales, Director of Health, ProPacífico; Dr. Marshall Summar, Director, Rare Disease Institute at Children's National, Washington, D.C. & Chairman of Board of Directors of the National Organization for Rare Disorders (NORD), USA; and Dr. Simon Kos, Chief Medical Officer and Senior Director, Worldwide Health, Microsoft & member of the Global Commission on ending the Diagnostic Odyssey of Children with Rare Diseases

Key asks included:

- » a) inclusion and participation of people living with a rare disease and their families in society;
- » b) well-being and optimal health;
- » c) improvement of health and social outcomes with the appropriate care and support within existing resources;
- » d) introduction of rare diseases into UN agencies and programmes, promotion of national strategies and actions, and regular reports by the UN Secretariat to monitor the progress on the implementation.



What you are doing here is helping the Member States to implement the 17 goals they agreed upon »



Daniela Bas, Director of Division for Inclusive Social Development, UN Department of Economic and Social Affairs (DESA), United Nations Secretariat



The French Third National Plan for rare diseases is based on a very strong principle: the inclusion and participation of people living with a rare disease in the conception and the implementation of said plan. It needs to be a concerted effort of all stakeholders »

Hayet Zeggar, Counsellor Health and Social issues, Permanent Mission of France to the United Nations

Durhane Wong-Rieger, Chair of the Council of RDI, made clear that the rare disease community is not coming to the United Nations empty-handed. It is coming 'with tools, plans, strategies' and most of all, 'with a lot of people who are personally very committed'.

The hope is to work towards this UN Resolution in the coming years, with the maximum support from Member States and international institutions. Notable speakers in the audience already issued statements amenable to this process.



Durhane Wong-Rieger, President and CEO, Canadian Organization for Rare Disorders (CORD); Chair of the Council, Rare Diseases International (RDI)



The UAE fully supports elevating the issue of rare diseases in the UN Agenda. We all heard the call for a UN resolution on rare diseases and, as a Member State, we could ask the experts to give us something to work on. We should not only encourage the national level policies but also, and more importantly, the UN system must not leave behind people with rare diseases »

Saud Al Shamsi, Deputy Permanent Representative of the United Arab Emirates to the United Nations



We have taken the motto "Leave no one behind" to heart at the national level, and as a country, we are in favour of the policy directions you are pursuing at the United Nations »

Tareq Albanai, Counsellor at Permanent Mission of the State of Kuwait to the United Nations



Malta stands ready with you, and with all Member States, to motivate others to come on board and work for this noble cause so that truly no one will be left behind »

H.E. Carmelo Inguanez, Ambassador and Permanent Representative of Malta to the United Nations

➔
Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe; Member of Council and Chair of Advocacy Committee, Rare Diseases International (RDI) launches a call for a UN General Assembly Resolution on Rare Diseases



Next steps towards the global empowerment of persons living with a rare disease [as of April 2019]

Following 2019 Rare Disease Day Policy Event at the United Nations, the NGO Committee for Rare Diseases vowed to work on two main priorities in the time to come: the inclusion of rare diseases in strategies towards Universal Health Coverage (UHC) and the adoption of a UN General Assembly Resolution on Addressing Holistically the Challenges of People Living with a Rare Disease.

Due to momentum and specific timing, the focus in 2019 will be on advocating towards the inclusion of rare diseases within UHC, in particular in the Political Declaration on UHC to be discussed and endorsed by all 193 Member States ahead of the High-Level Meeting on UHC on 23rd September 2019. Rare Diseases International (RDI) has released in April 2019 its position paper "Rare Diseases in Universal Health Coverage to Leave No One Behind". In addition, considering UHC is also a top priority for the World Health Organisation and will be the central focus of the 72nd World Health Assembly in May, the NGO Committee for Rare Diseases will support its members

and collaborators in the organisation of side-events on the topic of rare diseases at the WHA in Geneva. On the 23rd May 2019, a formal event within the WHA will be organised by EU Delegation, Romania and Kuwait, co-sponsored by several other Member States, with RDI on "How transformational digital technology can contribute to Universal Health Coverage: the case of rare diseases". Also on 23rd May, RDI and the NGO Committee for Rare Diseases will organise an Informal Side Event titled "Universal Health Coverage: Including Rare Diseases to leave no one behind" at the International Red Cross HQ.

In parallel, the plan of action is to take targeted incremental steps towards the durable inclusion of rare diseases in the UN 2030 Agenda with the goal of obtaining a Resolution that would ask governments and UN bodies and agencies to take into account the special and urgent needs of the particularly vulnerable population of persons affected by a rare disease.



Rare Disease Day Policy Event attendees outside the United Nations Headquarters in New York

The process leading to the adoption of a GA resolution on rare diseases must be planned in the long-term. Firstly, it involves a process of consultation in order to develop a draft that accurately represents the needs of the members of the community. Secondly, in parallel, the NGO Committee for Rare Diseases and its members and collaborators must catalyse and mobilise the support from Member States. A UN GA resolution requires the support of at least one UN Member State but has more strength if a group of co-sponsors from different regions of the world table the draft together.

To ensure legitimacy and a needs-centred approach, Rare Diseases International, member of the NGO Committee for Rare Diseases, will lead an internal consultation with its members to ensure people living with a rare disease themselves are involved in the process towards the Resolution on Rare Diseases. In parallel, the NGO Committee for Rare Diseases will facilitate the integration of the expert view points from all stakeholders involved in the field. The backbone of the Resolution will come from

elements like the 'RDI Joint Declaration', a set of twelve main recommendations to be addressed at international level as a matter of priority, as well as the outcomes of the two high level meeting organised by the NGO Committee for Rare Diseases. The sponsoring Member States will actively draft the resolution based on these civil society and expert contributions presented to them through the NGO Committee for Rare Diseases.

By taking this system-strengthening approach, the NGO Committee for Rare Diseases aims to create public awareness on the issues faced by people with a rare disease while urging governments and the United Nations system to include rare diseases in their policies and budgets for the long-term.

Although the challenges continue to be many and much remains to be done on an international level, the rare disease community is strong and ready to make its voice heard globally. 'Leave no one behind' means leaving no person living with a rare disease behind.



Rare Disease Day Policy Event Cocktail Reception, kindly hosted by the Ambassador and Permanent Representative of Spain to the United Nations, H.E. Agustin Santos Maraver

Rare Disease Day Policy Event Programme

Morning Plenary: 9:45 to 13:30

KEYNOTE ADDRESSES

9:45-11:00

LEAVE NO PERSON LIVING WITH A RARE DISEASE BEHIND

- 1 The NGO Committee for Rare Diseases: working on a common cause at the global level**
Mr. Anders Olason, Chair of the NGO Committee for Rare Diseases; Chairman of Ågrenska Foundation
- 2 Remarks by the Host**
H.E. Mr. Sven Jürgenson, Ambassador and Permanent Representative, Permanent Mission of Estonia to the United Nations
- 3 Rare Diseases and the Sustainable Development Goals**
Ms. Marion Barthelemy, Director, Office of Intergovernmental Support and Coordination for Sustainable Development, Economic and Social Council (ECOSOC)
- 4 Human Rights of people living with a rare disease**
Mr. Andrew Gilmour, Assistant Secretary-General for Human Rights and Head of the Office of the High Commissioner for Human Rights in New York
- 5 Remarks by Chair of the UN Group of Friends of Universal Health Coverage**
H.E. Toshiya Hoshino, Ambassador and Deputy Permanent Representative, Permanent Mission of Japan to the United Nations
- 6 Universal Health Coverage and Rare Diseases knowledge improvement: a journey to leave no one behind**
Ms. Pilar Aparicio, Director General of Public Health, Quality and Innovation at the Ministry of Health, Consumption and Social Welfare of Spain
- 7 Universal Health Coverage for people living with a rare disease**
H.E. Vitavas Srivihok, Ambassador and Permanent Representative, Permanent Mission of Thailand to the United Nations

This session aims to give a clear overall view of the developments that the rare disease community has made in the last two decades (in public awareness, healthcare systems, support systems and in research), with the particular goal of demonstrating the **need to consider rare diseases as a collective grouping** rather than looking solely at each individual disease. As people living with a rare disease face a number of **common needs and challenges**, they demand a **specific holistic strategy** and are an example of **traditionally disadvantaged social groups**, with the added specificity of the 'rarity'. This clearly demonstrate the **added-value of collaborating across diseases/conditions, and across countries**.

Co-Chairs:

- + **Mr. Terkel Andersen**, President of the Board of Directors, EURORDIS-Rare Diseases Europe
- + **Ms. Mandeep Dhaliwal**, Director of the HIV, Health and Development Group in the Bureau of Policy and Programme Support at United Nations Development Programme (UNDP)

- 8 Raising awareness in society at the global level**
Ms. Avril Daly, Chief Executive Officer, Retina International, Vice-President of the Board of Directors, EURORDIS-Rare Diseases Europe
- 9 Giving existence to people living with a rare disease in health systems**
Ms. Ana Rath, Director, Orphanet
- 10 A need for visibility within support systems**
Mr. Lieven Bauwens, Secretary General, International Federation for Spina Bifida and Hydrocephalus; Member of Inception Executive Board NGO Committee for Rare Diseases
- 11 Inclusion of rare diseases in scientific research**
Dr. Irene Norstedt, Acting Director responsible for the Health Directorate within the Directorate-General for Research and Innovation, European Commission and Head of Unit, Innovative and Personalised Medicine Unit; Member of the International Rare Diseases Research Consortium (IRDiRC)

SESSION 2

11:00-12:00

RECOGNISING RARE DISEASES AS A POLICY PRIORITY: EMPOWERING PEOPLE LIVING WITH A RARE DISEASE

Comfort Break

SESSION 3

12:15-13:30

UNIVERSAL
HEALTH
COVERAGE AND
REDUCTION OF
INEQUALITIES
FOR INCLUSIVE
DEVELOPMENT

This session aims to highlight policy priorities for persons living with a rare disease that are essential for the achievement of **Universal Health Coverage (UHC) and efficient and equitable health care systems**. Persons affected often suffer from **marginalisation and pauperisation** due to limited or scattered expertise, lack of diagnosis or misdiagnosis, and disproportionate out-of-pocket spending on health. But in addition, people living with a rare disease are often lost in the system, having to visit different health, social and local services in a short period of time and interacting with actors that work in silos. Throughout the sub-sessions, speakers will demonstrate the need for and the benefits arising from policies related to diagnostic, cross-border and cross-sector collaboration, and use of emerging technologies. Sub-session 3 will particularly highlight the **importance of thinking beyond health systems** solely and promoting measures that are **multidisciplinary, holistic, continuous, person-centred and participative** in nature. Such policies will significantly reduce the burden on everyday life, prevent discrimination and stigma, and contribute to the **enjoyment of fundamental rights** and to the fulfilment of the **full potential** of persons living with a rare disease as pledged by Member States under the UN **2030 SDGs agenda**.

Co-Chairs:

- + **Dr. Nata Menabde**, World Health Organisation NYC Office Director
- + **Mr. Alain Weill**, President of World Hemophilia Federation and member of board of NGO Committee for Rare Diseases

3.1 DIAGNOSIS AS A DOOR-OPENER

12 The extreme of 'Leave no one behind' – undiagnosed patients

Dr. William Gahl, Chair, Undiagnosed Diseases Network International (UDNI); Clinical Director, National Institutes of Health, National Human Genome Research Institute

13 New opportunities to improve the diagnosis of children living with a rare disease

Dr. Simon Kos, Chief Medical Officer and Senior Director, Worldwide Health, Microsoft & member of the Global Commission on ending the Diagnostic Odyssey of Children with Rare Diseases

3.2 LOCAL HEALTHCARE PROVISION AND GLOBAL NETWORKING

14 Rare Diseases as an opportunity for global collaboration

Dr. Ruediger Krech, Director, Universal Health Coverage and Health Systems, World Health Organisation

15 Case-Study: European Reference Networks

Mr. Martin Seychell, Deputy Director-General for Health and Food Safety, DG SANTE, European Commission

3.3 HOLISTIC APPROACH: BRIDGING HEALTH AND SOCIAL CARE

16 The need to address the 'Big 5' from a lifelong perspective: healthcare, social care, school, insurance and labour

Mr. Robert Hejdenberg, Chief Executive Officer, Ågrenska Foundation

Lunch Break 13:30 to 14:30

Buffet to be served by the stairs behind the left side of the Vienna Café



SESSION 4

14:30-17:15

ENSURING
INCLUSION AND
PARTICIPATION
OF PEOPLE LIVING
WITH A RARE
DISEASE THROUGH
NATIONAL POLICIES
AND STRATEGIES

This session aims to showcase **national strategies, case-studies and pilots** put in place in a number of Member States and carried **in collaboration with organisations of people living with a rare disease and/or with government support**. These illustrate the benefits that ensue when specific national strategies for rare diseases are established and how they contribute towards inclusive and equitable societies.

Co-Chairs:

- + **Ms. Daniela Bas**, Director of Division for Inclusive Social Development, UN Department of Economic and Social Affairs (DESA), United Nations Secretariat
- + **Ms. Durhane Wong-Rieger**, President and CEO, Canadian Organization for Rare Disorders (CORD); Chair of the Council, Rare Diseases International (RDI)

4.1 NATIONAL COMPREHENSIVE STRATEGIES

- 17 **Nan-Byo Strategy – Japan**
Mr. Toshi Ezoe, Counsellor, Permanent Mission of Japan to the United Nations
- 18 **National plan for Rare diseases - France**
- 19 **Implementing a national rare disease plan starting with national registry– Colombia**
Mr. Germán Escobar Morales, Director of Health, ProPacífico
- 20 **Establishing an APEC Rare Disease Registry Network: QUT and a framework for collaboration**
Prof. Matthew Bellgard, Director of eResearch, Division of Research and Innovation, Queensland University of Technology and Chair of APEC LSIF Rare Disease Network

4.2 INTEGRATED APPROACH BETWEEN RESEARCH AND CARE; AND BETWEEN MEDICAL AND SOCIAL CARE

- 21 **Networks for applying research to diagnosis and care – Canada**
Dr. Kym Boycott, Professor of Pediatrics, University of Ottawa; Care4Rare
- 22 **Rare Diseases Clinical Research Networks – United States of America**
Dr. Marshall Summar, Director, Rare Disease Institute at Children's National, Washington, D.C; Chairman of Board of Directors of the National Organization for Rare Disorders (NORD), USA
- 23 **Resource centres for rare diseases – Romania**
Ms. Dorica Dan, President, RONARD (Romanian National Alliance for Rare Diseases); Member of Board, EURORDIS-Rare Diseases Europe

Comfort Break

4.3 COUNTRIES EMERGING TO THE CHALLENGES OF RARE DISEASES: FROM THE GRASSROOTS LEVEL TO THE POLICY LEVEL

- 24 **Building a grassroots approach for persons living with a rare disease – Iran**
Dr. Hamid. R. Edraki, Managing Director, Rare Diseases Foundation of Iran (RADOIR)
- 25 **The case of rare diseases in China**
Dr. Shuyang Zhang, Vice President of Peking Union Medical College Hospital (PUMCH), Director of Clinical Pharmacology Research Center, PUMCH
- 26 **The case of rare diseases in Brazil**
Mr. Ricardo Monteiro, Minister-Counsellor, Permanent Mission of Brazil to the United Nations
- 27 **Integration of rare diseases in the national health system - Philippines**
Ms. Cynthia Magdaraog, President of the Philippine Society for Orphan Disorders, Inc. (PSOD)
- 28 **Strategies for rare diseases within government's commitment to healthcare - South Africa**
Ms. Kelly du Plessis, Chief Executive Officer, Rare Diseases South Africa
- 29 **Regional collaboration for care of people living with a rare disease – Kuwait**
Mr. Tareq Albanai, Counsellor at Permanent Mission of the State of Kuwait to the United Nations
- 30 **Call for action on rare diseases to the Members of the United Nations**
Mr. Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe; Member of Council and Chair of Advocacy Committee, Rare Diseases International (RDI)
- 31 **Closing**
Mr. Anders Olason, Chair of the NGO Committee for Rare Diseases; Chairman of Ågrenska Foundation
- 32 **Closing Keynote Addresses**
Official video statement from **Mrs. Tamara Vucic**, Spouse of the President of the Republic of Serbia.
Official video statement from **Mrs. Michelle Muscat**, Spouse of the Prime Minister of the Republic of Malta.
Mr. Anders Nordström, Ambassador Global Health, Swedish Ministry of Foreign Affairs
H.E. Mrs. Lana Z. Nusseibeh, Ambassador and Permanent Representative, Permanent Mission of the United Arab Emirates to the United Nations

CLOSING SESSION

17:45-18:15



A Substantive Committee of
The Conference of NGOs in Consultative Relationship
with the United Nations

The NGO Committee for Rare Diseases is a substantive committee established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO).

The NGO Committee for Rare Diseases was initiated by the Agrenska Foundation and EURORDIS-Rare Diseases Europe, with the later integration of Rare Diseases International (RDI), with a view to bringing greater political recognition of the challenges of rare diseases at the global level. Its formation was approved by a vote of 27 CoNGO member organisations in April 2014, and its official inauguration at the United Nations took place on 11 November 2016 in New York.



www.agrenska.se



www.eurordis.org



RARE
DISEASES
INTERNATIONAL

www.rarediseasesinternational.org



NGO COMMITTEE FOR
RARE DISEASES

ngocommitteerarediseases.org  [@ngorarediseases](https://twitter.com/ngorarediseases)