



RARE  
DISEASES  
INTERNATIONAL

# HOPES AND CHALLENGES OF PEOPLE LIVING WITH A RARE DISEASE WITHIN UNIVERSAL HEALTH COVERAGE

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International

# RARE DISEASES

OVER

**6000**

distinct rare diseases

Affects approx.



of the population in  
the course of their lives

Each one affects  
fewer than

**1 IN**

**2000** PEOPLE

**NO  
CURE**



for the vast majority  
of diseases and few  
treatments available



All together,  
an estimated

**25/30**  
**MILLION PEOPLE**

are living with a rare  
disease in Europe and

**300** MILLION  
worldwide



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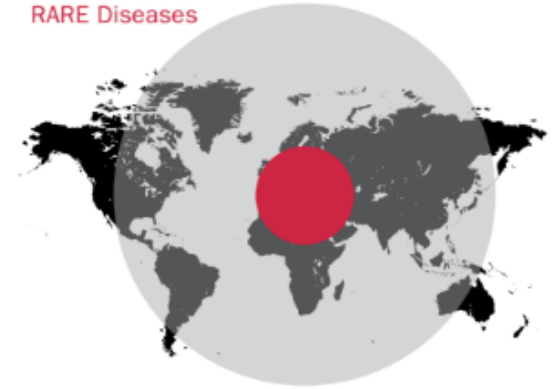
# RARITY CALLS FOR ACTION AT THE GLOBAL LEVEL

No one country, no one continent, can solve alone the problems posed by rare diseases

- Need to bring together a **critical mass** of patients and medical experts, scientists and public health authorities.
- Need to support countries emerging to rare diseases and promote **international collaboration** between researchers, medical experts and other stakeholders worldwide
- **Civil society empowerment** at international level

300 MIO / 7'052 MIO

RARE Diseases



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# THE CHALLENGES

Most rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening



- Patients and experts are few, geographically **scattered** and often **isolated**
- Patients are undiagnosed, misdiagnosed or wait years for a diagnosis. Highly heterogeneous diseases
- Reliable **information is scarce**
- **Fragmented research**, data and expertise. Lack of **treatments**: difficulties to access adequate care
- High social impact and **marginalisation** within society at large and within healthcare systems designed for common diseases
- **Heavy psychosocial burden**

A rare disease affects...



HEALTH



FAMILY



EDUCATION



WORK



ECONOMY



SOCIETY



SCIENCE



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# THE HOPES

- Commonalities of issues and critical mass enables common policies across diseases
- New opportunities in 21st century
- Scientific advances and translational research
- New technologies : digital, AI, 3D, nano...
- Global networking
- Translate into more and earlier diagnosis; better care and innovative treatments for patients; improved patient health outcomes and quality of life



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# JOINT DECLARATION OF RARE DISEASE PATIENTS

## “RARE DISEASES: AN INTERNATIONAL PUBLIC HEALTH PRIORITY”

The patient movement worldwide has adopted the following twelve main recommendations to be addressed at international level as a matter of priority:

### Visibility of Rare Diseases

1. Enhance visibility of rare diseases at an international level in terms of public awareness and within the healthcare system as a human rights issue and a public health priority.
2. Better classification and codification of rare diseases can support the process towards better recognition of rare diseases worldwide.
7. Facilitate universal access to high quality healthcare and treatments for rare diseases patients, including surgeries, special diets, transplants, and medical devices, common and orphan medicinal products.
8. Create policies on rare diseases that promote specific measures in regional / national / international strategies, including identification and support of specialised expert providers as well as their national and international networking.

### Patient Empowerment

3. Support and empower patients and families to play an active role in shaping national health care provision that is appropriate to their needs.
4. Develop, gather, share and disseminate information on rare diseases in linguistically and culturally appropriate formats.
5. Promote international cooperation in the field of services to patients and families.
9. Promote recognition that rarity requires increased international cooperation and mobility of experts as well as of patients when expertise is not available locally.

### Universal Access to Healthcare

6. Improve access to services that will facilitate informed decisions about prevention and screening that are legally permissible, and improve access to accurate and timely diagnoses.

### Research

10. Coordinate worldwide research efforts on rare diseases through international and national research initiatives.
11. Build opportunities for effective networking of patient registries.

### Cooperation in policy shaping at international level

12. Elaborate policies based on common values (equity, solidarity and Social Justice) that have a positive impact on the lives of rare disease patients.



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NGO COMMITTEE FOR  
**RARE DISEASES**

**11 November 2016**

**United Nations, New York**

# The Global Gathering for Rare Diseases

INAUGURATING THE NGO COMMITTEE  
FOR RARE DISEASES



# 2016: LAUNCH OF NGO COMMITTEE FOR RARE DISEASES AT THE UNITED NATIONS IN NEW YORK

November 11, 2016



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# A MILESTONE IN THE JOURNEY TO PUT RARE DISEASES IN THE GLOBAL AGENDA

## Founding Act

### « Rare Diseases and the UN Sustainable Development Goals »

New York City, Friday 11 November 2016

We, the undersigned participants to the inaugural « Global Gathering for Rare Diseases », proclaim and endorse the following statements:

- Each of the 6,000 repertoried rare diseases affects a very small population locally. **All together, however, they represent an international public health issue.**
- The 350 million persons living with a rare disease worldwide are facing **common challenges across diseases and across borders** that affect all parts of their lives at once.
- No one country, no one continent alone can solve the problems posed by rare diseases.**
- Experience shows that actions are possible and effective. **Common national policies and international collaboration can address these challenges.**
- Scientific, medical, technological, social opportunities will be high in the next 20 years. **Patients, NGOs and other stakeholders are committed and aligned for partnership.**
- Rare diseases fit within the objectives from several UN SDGs**  and can significantly contribute to their achievement.

The NGO Committee for Rare Diseases will catalyse all efforts towards the delivery of the SDGs in support of rare diseases, and towards the recognition and integration of rare diseases in all relevant future global policy and initiatives of the United Nations and its agencies.



NGO COMMITTEE FOR  
RARE DISEASES  
RARE DISEASES

Handwritten signatures of participants, including: Richard J. Gutter, Lehm, Michael, Hans, Anna, ALBA DE JONG, Hassan Van der, Kerkhof, Pien, and others.



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## SDGS

## RARE DISEASE CHALLENGES

## SDGS

## RARE DISEASE CHALLENGES

## 1 NO POVERTY



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

## 8 DECENT WORK AND ECONOMIC GROWTH



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

## 3 GOOD HEALTH AND WELL-BEING



Persons with a rare disease need more and better medicines, appropriate diagnosis and lifelong care and social support

## 9 INDUSTRY, INNOVATION AND INFRASTRUCTURE



There is a need to invest in research & development of therapies, health technologies and diagnostic tools

Accessibility to infrastructure is key to be included in society

## 4 QUALITY EDUCATION



50% of rare diseases affect children who often face difficulties attending school due to inaccessibility of facilities and non-adapted teaching methods

## 10 REDUCED INEQUALITIES



Persons with a rare disease tend to remain a marginalised population suffering from discrimination in the health, labour and governance fields

## 5 GENDER EQUALITY



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

## 17 PARTNERSHIPS FOR THE GOALS



The rare disease community is increasingly interconnected, with a myriad of networks of patient advocates, regulators, research & industry

# 2017: 1ST RARE DISEASE POLICY EVENT IN GENEVA



**THE RIGHT TO HEALTH:  
THE RARE DISEASE  
PERSPECTIVE**

Friday, February 10, 2017  
**#rdiGeneve**

In partnership and  
with the support of:

**BLACKSWAN  
FOUNDATION**  
INTERNATIONAL FOUNDATION FOR RARE DISEASES

**EURORDIS**  
RARE DISEASES EUROPE

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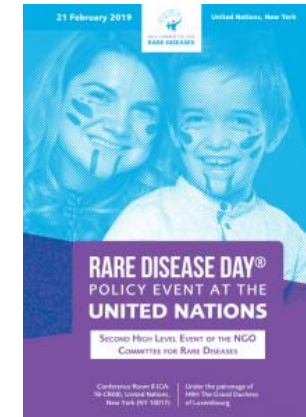


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# 2019: RARE DISEASE DAY POLICY EVENT AT THE UNITED NATIONS

- **Second High-Level Event of the NGO Committee for Rare Diseases, 21 February 2019, United Nations Headquarters, New York**



Under the patronage of

**HRH The Grand Duchess  
of Luxembourg**



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# 2019: RARE DISEASE DAY POLICY EVENT AT THE UNITED NATIONS



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# THE VOICE OF THE RARE DISEASE PATIENT COMMUNITY

## The Rare Disease Community Position



**Rare Diseases:**  
Leaving no one behind in Universal  
Health Coverage



RDI Position Paper released in April 2019

- RD and UHC movements share important commonalities in spirit and vision
- UHC shall never be fully attained nor realised if persons living with rare diseases are left behind and their needs left unmet.
- The time to integrate rare diseases in the reflection on, and practice of, Universal Health Coverage is now.



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**International Universal Health Coverage Day  
12 December 2018, United Nations Headquarters, New York**

RDI was invited to participate in a Discussion Panel organised by WHO and Group of Friends of UHC (Japan, Brazil, France, Ghana, Hungary, South Africa, Thailand) at United Nations in New York. RDI was the only civil society organisation represented in the panel.

Durhane Wong-Rieger, Chair of the Council of RDI addressed dignitaries and delegates.



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# KEY ASKS FROM THE UHC MOVEMENT FOR THE UN HIGH-LEVEL MEETING ON UHC

- ASK 1: Ensure political leadership beyond health – Commit to achieve UHC for healthy lives and wellbeing for all at all stages, as a social contract.
- ASK 2: Leave no one behind – Pursue equity in access to quality health services with financial protection.
- ASK 3: Regulate and legislate – Create a strong, enabling regulatory and legal environment responsive to people's needs
- ASK 4: Uphold quality of care – Build quality health systems that people and communities trust.
- ASK 5: Invest more, invest better – Sustain public financing and harmonize health investments.
- ASK 6: Move together – Establish multi-stakeholder mechanisms for engaging the whole of society for a healthier world.



THANK YOU!



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