Rare Diseases International
2019 Activity Report

RDI is a global alliance of people living with a rare disease of all nationalities across all rare diseases. RDI brings together rare disease patient organisations from around the world as well as international rare disease-specific federations to create the global alliance of rare disease patients and families.

RDI was launched in 2015 as a EURORDIS Initiative, and incorporated as a separate legally-registered organisation as of 2019, to create a strong common voice on behalf of all people living with a rare disease around the world, to advocate for rare diseases as an international public health priority, to represent its members, and to enhance their capacities.

The objectives of RDI are to:
- Unite, expand and reinforce the movement of people living with a rare disease to speak with one strong voice.
- Establish rare diseases as a public health priority in more countries and regions around the world as well as at the global level.
- Put rare diseases on the agenda of the United Nations and other international organisations.
- Strengthen rare disease patient groups’ capacity to act at local, national, regional and global levels.

Milestones in 2019

Governance

RDI incorporated in 2019

RDI was incorporated under French law as an Association Loi 1901 on December 7, 2018 and has been operating as a legally-registered organisation since January 2019.

The Statutes (constitution) of the organisation can be found on the website: https://www.rarediseasesinternational.org/governance/

By-Laws (internal rules) are under development for adoption in April 2020.

Council Elections

RDI’s Council is elected by full members and ensures a credible and effective governance of RDI. It also advises on the programme’s long term strategy and actions.
Since the 2019 General Assembly took place early in the year (February 2019), Council elections had to be organised at the end of 2018. Therefore, in December 2018, all full members of RDI had the opportunity to vote online to replace or re-elect the following outgoing members of the Council:

- Yann Le Cam, EURORDIS- Rare Diseases Europe (term 2016 – 2019)
- Durhane Wong-Rieger, Canadian Organization for Rare Disorders (term 2016 – 2019)
- Alfredo Toledo, Alianza Iberoamericana de Enfermedades Raras (term 2017- 2020) who stepped down in September 2018
- Angela Chaves Restrepo, Federación Colombiana de Enfermedades Raras (term 2018- 2021) who stepped down in July 2018

<table>
<thead>
<tr>
<th>Name of representative</th>
<th>Organisation</th>
<th>Year of Election</th>
<th>End of mandate</th>
</tr>
</thead>
<tbody>
<tr>
<td>KP Tsang</td>
<td>Retina International</td>
<td>2017</td>
<td>2020</td>
</tr>
<tr>
<td>Jesus Navarro</td>
<td>ALIBER - Iberoamerican RD Alliance</td>
<td>2019</td>
<td>2020</td>
</tr>
<tr>
<td>Durhane Wong-Rieger</td>
<td>Canadian Organization for Rare Disorder</td>
<td>2019</td>
<td>2022</td>
</tr>
<tr>
<td>Yann Le Cam</td>
<td>EURORDIS - Rare Diseases Europe</td>
<td>2019</td>
<td>2022</td>
</tr>
<tr>
<td>Rachel Yang</td>
<td>Chinese Organization for Rare Disorders</td>
<td>2018</td>
<td>2021</td>
</tr>
<tr>
<td>Lisa Phelps</td>
<td>NORD</td>
<td>2018</td>
<td>2021</td>
</tr>
<tr>
<td>Ritu Jain</td>
<td>Debra International</td>
<td>2018</td>
<td>2021</td>
</tr>
</tbody>
</table>

Yann Le Cam was re-elected with 20 votes and Durhane Wong-Rieger of Canadian Organization of Rare Disorders was r-elected with 20 votes for another 3-year mandate.
Rachel Yang from Chinese Organization for Rare Disorders, with 13 votes, and Jesús Navarro for the Alianza Iberoamericana de Enfermedades Raras, with 11 votes, were elected to replace Angela Chaves (FECOER) and Alfredo Toledo (ALIBER) for the remainder of their terms.

A total of 8 nominations were received and 21 full members voted.

**A growing movement of people living with a rare disease**

**Membership**

An engaged membership base is key to establish an effective global alliance of people living with a rare disease. From starting out with 20 member organisations in 2015, RDI had 62 member organisations at the end of 2019. Member umbrella organisations represent patient groups from 35 national alliances, 3 pan regional networks and 17 international disease-specific federations. Through RDI members, rare disease patients are represented in more than 100 countries worldwide.

The following rare disease patient umbrella organisations joined RDI in 2019:

1. Instituto Vidas Raras (Brazil)
2. Pan Cyprian Federation of Patients’ Associations and Friends
3. Rare Disease Ghana Initiative
4. ESPERANTRA (Peru)

The list of RDI members is continually updated and published online at: [rarediseasesinternational.org/members](http://rarediseasesinternational.org/members)

**Communication & Information Sharing**

In 2019, RDI members started receiving an electronic monthly newsletter with a round-up of news and events pertaining to RDI and the international rare disease community.

In September, RDI launched its own Facebook Workplace, a virtual space for members to connect and exchange information. Since launch, 36 representatives from member organisations have joined to share their news and events, to exchange information with peers, ask questions on cross-cutting topics and learn from other patient advocates working at national, regional and international level.

The RDI website ([www.rarediseasesinternational.org](http://www.rarediseasesinternational.org)) is continually been updated and is the main source of information for the general public on what is on the agenda of the patient rare disease global movement.
RDI’s 5th Annual Meeting, New York, USA

RDI’s 5th Annual Meeting took place on February 20th, 2019 at the Microsoft Global Headquarters in New York, USA back to back to the Launch of the Recommendations Report of the Global Commission to End the Diagnosis Odyssey of Children with a Rare Disease and the day before the NGO Committee for Rare Diseases High Level Event which was on the occasion of Rare Disease Day at the United Nations Headquarters in New York.

40 delegates from 32 member organisations from 26 countries attended the General Assembly and Membership Meeting (closed meeting for members only).

119 delegates registered to attend the RDI Global Meeting, which was open to member and non-member patient groups, as well as all other stakeholders. As the meeting took place for the first time outside Europe, the meeting was also well attended by US-based patient organisations and NGOs in consultative relations with the UN New York.

Building a community of global patient advocates

RDI’s Advocacy Committee

Yann Le Cam, RDI Council Member (EURORDIS – Rare Diseases Europe) is the Chair of the Committee. 16 patient representatives from RDI’s member organisations, plus a representative from the International Alliance of Patient Organisations (IAPO), take part in this Committee and represent a wide range of countries and diseases.

They following patient advocates joined the Advocacy Committee in 2019:

- Eleni Anoniou, TIF
- Roberta Anido, FADEPOF
- Luciana Escaì, FADEPOF
- Richards, WFH
- Leire Solis, IPOPI
The following patient advocates left the Advocacy Committee in 2019

- Angela Chaves FECOER
- Paul Melmeyer NORD
- Mark Brooker WFH
- Chiara Ciriminna BLACKSWAN Foundation

**Partnering with other sectors to build a sustainable organisation**

**RDI Alliance of Companies for Patient-Centred Action**

Launching of a platform for companies to support the rare disease cause internationally and to increase mutual understanding of issues that are important for rare disease patients and different industries.

The Alliance of Companies aims to:

- Build an international, cross-sector network of companies working in areas that affect rare disease patients. Examples include pharmaceutical and biotechnology, medical equipment and devices, research, information technology, data mining and management, artificial intelligence, transport and logistics.
- Exchange perspectives on issues vital to rare disease patients
- Stimulate companies across many industries to address patient needs
- Survey results, key findings, member feedback on patient needs
- Provide financial support to advance RDI’s mission

In 2019, the following 9 companies joined the Alliance: Actelion, Bridgebio, CSL Behring, Kyowa Kirin, Pfizer, Pulse Infoframe, Sanofi Genzyme, Takeda and Vertex.

Representatives from 7 companies in the Alliance attended the open session of the 5th RDI Annual Meeting at Microsoft’s Global Headquarters in New York, and the Rare Disease Day event at UN Headquarters in New York on February 20-21st. Representatives from 3 companies in the Alliance attended the First Informal WHA Side Event on Rare Diseases on May 23rd in Geneva. On August 29, the members of the Alliance met together for the first
time at an online meeting organised by the RDI Secretariat to present them with RDI’s advocacy strategy and to discuss the impact on patients and companies of the inclusion of rare diseases in the UN Political Declaration on Universal Health Coverage.

Advocacy actions in the United Nations system

Rare Disease Day Policy Event at the United Nations

On 21 February 2019, RDI co-organised with the NGO Committee for Rare Diseases and EURORDIS – Rare Diseases Europe a Rare Disease Day Policy Event at the United Nations (the Second High-Level Event of the NGO Committee for Rare Diseases). The event, which was hosted by the Permanent Mission of Estonia to the UN, and co-hosted by 14 other Permanent Missions to the UN, was an opportunity to elevate rare diseases within the UN 2030 Sustainable Development Agenda.

Over 100 participants from the international NGO community, UN agencies, national governments, academic institutions, the private sector and the rare disease patient community came together at the United Nations Headquarters in New York.

40 patient advocates from RDI member organisations were invited to participate and represent the rare disease patient community.

At the event, Yann Le Cam, Chief Executive Officer of EURORDIS, launched a call for rare diseases to be included in universal health coverage (UHC) and also called on UN Member States to support a UN resolution on rare diseases. There was an overwhelming sense of support from Member States and UN and WHO speakers present.
Rare Diseases & Universal Health Coverage

RDI Position Paper on UHC

In April 2019 RDI adopted and released the Position paper ‘Rare Diseases: Leaving No One Behind in Universal Health Coverage’. The paper, which included input and featured country case studies from RDI members, argues that UHC shall never be attained or realized if persons living with rare diseases are left behind and their needs left unmet.

The paper was instrumental in RDI’s campaign to enable rare disease alliances to advocate for the inclusion of rare diseases in the UN Political Declaration on UHC and will continue to serve to advocate for implementation of the commitments made within the Declaration - at both global and national level.

Paper available on RDI website: https://www.rarediseasesinternational.org/policy-positions/

Rare Diseases highlighted at the 72nd World Health Assembly

On May 23rd 2019, RDI organised two events on rare diseases in the context of the WHO 72nd World Health Assembly, gaining further momentum towards formal relations with WHO and the inclusion of Rare Diseases in the UN Political Declaration on UHC.

First Formal WHA Side Event on Rare Diseases

The formal side event: «How transformational digital technologies can contribute to leave no one behind in UHC: the case of rare diseases » was held inside the Palais des Nations, at the UN Headquarters in Geneva, and was part of the official WHA agenda. The event was sponsored by EU, Romania, Kuwait and was co-sponsored by an additional nine Member States.
Key speakers included: Dr Soumya Swaminatha, WHO Chief Scientist and Martin Seychell, Deputy Director-General DG SANTE, European Commission.

Prior to this event, RDI contributed to the WHO’s thinking on the topic of digital health by responding to the public consultation on the WHO Draft Global Strategy on Digital Health in April 2019.

First Informal WHA Side Event on Rare Diseases

The same day, RDI in collaboration with the NGO Committee for Rare Diseases, hosted an informal rare disease flagship Side Event at the International Museum of the Red Cross, on the sidelines of the WHA. The meeting under the theme “UHC: Including rare diseases to leave no one behind” gave the opportunity to present the position of RDI and show that Integration of rare diseases in policies and legislations is possible in all countries whatever their level of development (ex. Philippines)

The event was attended by 65 representatives of Member States and the rare disease community and was endorsed by the Permanent Mission to the UN of France, Georgia, Malta and the Netherlands. Key speakers included: Dr Rüdiger Krech, Director, Universal Health Coverage and Health Systems, Office of the Assistant Director-General, World Health Organization and Todd Howland, Chief of the Development and Economic and Social Issues Branch, Office of the United Nations High Commissioner for Human Rights.

Rare diseases recognised by UN Human Rights

In July rare disease was included in the 2019 Annual Report of the Office of the High Commissioner for Human Rights (OHCHR) “The Right to Health: how human rights framework can contribute to implementation of UHC.” The report refers to persons living with a rare disease as a particular focus group to take into account when implementing UHC
within a human rights framework (paragraphs. 28, 29, 47 and recommendation 51). The report references RDI Position Paper on UHC.

**Rare Diseases included in landmark UN Political Declaration on Universal Health Care**

In July-August 2019, RDI and EURORDIS led a joint campaign with 13 national rare disease alliances to reach out to national ministries of health and foreign affairs; and to Permanent Missions to the UN – to advocate for the inclusion of rare diseases in the final text of the UN Political Declaration on UHC.

On September 23, 2019 the UN Political Declaration on UHC was adopted by 193 Member States and endorsed by all Heads of State at High-Level Meeting on UHC during UNGA 74th Session. The Declaration includes rare diseases (Article 34) and engraves it as an area that needs to be addressed as part of UHC.

RDI Council members, Durhane Wong Rieger and Yann Le Cam represented the rare disease patient community at the UN High Level Meeting in New York and submitted a written statement which will remain as part of the official proceedings.

In November, RDI was formally accepted as a member of UHC2030, the global movement to build stronger health systems for universal health coverage led by the WHO and the World Bank. RDI will use this and other platforms to hold governments accountable of their commitments towards rare diseases and UHC.

**Start of formal collaboration with World Health Organization**

On 18 March 2019, Yann Le Cam and Durhane Wong-Rieger met with Dr. Tedros Adhanom Ghebreyesus, Director-General of the WHO, in Geneva. The meeting was a highlight in the journey to carve a space for rare disease within the WHO programme of work and served to identify areas of collaboration in view a Memorandum of Understanding between the two organisations.

In December, RDI and WHO signed a Memorandum of Understanding based on ambitious goals and long-term framework of collaboration that will contribute to WHO’s 13th General Programme of Work.

In the first year, activities under the Memorandum of Understanding (MoU) will focus on a framework methodology to define rare diseases internationally and on laying the ground for the
development of a global network of centres of excellence for rare diseases.

Supporting countries emerging to rare diseases and building capacities amongst patient leaders of the rare disease community

Conference & Fellowship Programmes

As part of the Conference Programme RDI supported the following conferences:

- *Rare Disease Day at the UN Event, Global Commission Report Launch, New York, Feb 20-21 level 3*
  - RDI financial support for patient representatives to participate (3 speakers and 14 fellows)
  - *World Orphan Drug Congress, Washington DC, April level 2*
  - *Canadian Organization for Rare Disorders/ ReACT Conference, Toronto, Canada, May 9-13 level 3*
  - RDI financial support for patient representatives to participate (4 speakers and 5 fellows)
  - ‘*Rare Diseases: A global challenge, a holistic challenge*’ *Conference at the National Rare Disease Centre (CREER), Burgos, Spain, September 19-20 level 2*
  - *APARDO Summit ‘Regional Collaboration for Global Change’, Taipei, Taiwan, October 19-21 level 3*
  - RDI financial support for patient representatives to participate (3 speakers and 13 fellows)
  - *XIV International Conference on Rare Diseases and Orphan Drugs” (ICORD), Tel Aviv, Israel, November 11-13 level 2*
  - *World Orphan Drug Congress, Barcelona, Spain, November 12-14 level 2*
  - *VI Encuentro Iberoamericano de Enfermedades Raras (ALIBER 6th Conference) Murcia, Spain, November 13-15 level 1*

The fellowship Programme was supported by Amicus Therapeutics and CSL Behring
Fostering dialogue with industry

**RDI-IFPMA Round Table on access for rare disease therapies in LMICs**

RDI co-organised with the International Federation of Pharmaceutical Manufacturers & Associations (IFPMA) a Round Table on access for rare disease therapies in lower and middle-income country settings. The meeting, which was held at the International Union for Cancer Control in Geneva on December 4th 2019, brought together 15 patient representatives from RDI’s network and 14 company representatives from the biopharmaceutical industry to explore barriers to access, discuss opportunities to overcome these challenges and identify what factors have helped to address them.

Patient advocates from Mexico, Guatemala, Uganda, Malaysia, US, UK, France, Cyprus and Portugal from disease areas including: haemophilia, SMA, thalassemia, rare cancers, Gaucher and other lysosomal storage disorders, described the expanded programmes that exist for their diseases at international level and more specifically about how they are carried out in their countries and the obstacles they encounter on the ground.

**RDI presence in the international rare disease ecosystem**

**The NGO Committee for Rare Diseases**

The NGO Committee for Rare Diseases (New York) is a substantive committee established under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO), since November 2016. The main objective of the NGO Committee for Rare Diseases is to bring visibility and understanding about rare diseases to the United Nations and align rare diseases with the UN 2030 Sustainable Development Agenda.
RDI’s Chair, Durhane Wong-Rieger is RDI’s representative at the NGO Committee for Rare Diseases.

RDI is a Member of the Inception Executive Board of the NGO Committee for Rare Diseases. The Chair of the Committee is Anders Olauson, Founder and Chair of the Agrenska Centre for Rare Diseases. The other members of the Executive Board are: EURORDIS, World Federation of Hemophilia, International Federation for Spina Bifida and Hydrocephalus, International Alliance of Patients’ Organisations and the International Alliance of Women.

**International Rare Disease Research Consortium (IRDIRC)**

Ritu Jain of DEBRA International represented RDI in IRDiRC’s Consortium Assembly and Patient Advocates Constituency Committee (PACC).

- The PACC is composed of 15 umbrella patient groups of which 13 are member organisations of RDI.

In 2019 Ritu Jain participated in a number of teleconferences of IRDIRC PACC and two face-to-face meetings in Leiden, Netherlands, May 22-23 and in Paris, France, November 21-22

**Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease**

The Global Commission is a high-level expert panel established in 2018 and led by Shire, Microsoft and EURORDIS with the purpose to build road map to improve diagnostic pathways for children with rare diseases.

- The following RDI patient representatives are members of the Global Commission: Yann Le Cam (EURORDIS), Durhane Wong-Rieger (CORD Canada), Kevin Huang (CORD China), Pamela Gavin (NORD)

In 2019 RDI members were invited to give their input on the Global Commission’s first-year recommendations and pilots; and participated in the launch of Year-1 Report at events in New York, Brussels and Beijing.

RDI Chair, Durhane Wong-Rieger, participated alongside Takeda’s Centre for Scientific Leadership and Innovation and other key stakeholders, in a Global Commission Workshop at IDEO in Cambridge, MA to discuss Patient Empowerment, and opportunities to move forward on Track 1 recommendations to equip families with information and tools to collaborate effectively with their primary care doctor to reach a diagnosis faster.

**APEC Rare Disease Network**
➢ APEC Rare Disease Policy Dialogue focused on Latin America, Santiago, Chile, July 10-11

In July, RDI Chair, Durhane Wong-Rieger was invited by the Asia-Pacific Economic Cooperation (APEC) Life Sciences Innovation Forum (LSIF) Rare Disease Network to a Meeting in Chile to engage with the patient community in the implementation of the APEC Rare Disease Action Plan. The Action Plan calls for APEC’s 21-member economies to improve the economic and social inclusion of individuals living with rare diseases, with clear targets by 2025.

➢ APEC Workshop ‘Using APEC Rare Disease Framework to develop National and Local Action’, Taipei, Taiwan, October 21

In October, RDI patient representatives from Asia and Ibero America attended a workshop hosted by the APEC Rare Disease Network in Taipei, back to back to APARDO Membership Summit and Taiwan Foundation for Rare Disorders 20th Anniversary celebrations.
Members

Alliance Maladies Rares
Allianz Chronischer Seltener Erkrankungen
Arabic Organisation for Rare Diseases
Asia Pacific Alliance of Rare Disease Organisations
Associacao Brasileira de Enfermedades Raras
Asociacion de Familiares y Afectados de Lipodistrofias
Association ANNA
Associação Portuguesa CDG
Blackswan Foundation
Botswana Organisation for Rare Diseases
Canadian Organization for Rare Disorders
Chinese Organization for Rare Disorders
Croatian Alliance for Rare Diseases
Cutix Laxa International
Cyprus Alliance for Rare Disorders
DEBRA International
ESPERANTRA Peru
EURORDIS-Rare Diseases Europe
Federación Argentina de Enfermedades Poco Frecuentes
Federación Colombiana de Enfermedades Raras
Federación Española de Enfermedades Raras
Federacion Mexicana de Enfermedades Raras
Federation of European Associations of Patients Affected by Renal Genetic Diseases
Findacure
Genetic Alliance Australia
Genetic Alliance South Africa
Greek Alliance for Rare Diseases
Hong Kong Alliance for Rare Diseases
Iberoamerican Alliance for Rare Diseases
Instituto Vidas Raras
Indian Organization for Rare Diseases
International Federation for Spina Bifida and Hydrocephalus
International Gaucher Alliance
International Niemann-Pick Disease Alliance
International Organisation for Primary Immunodeficiencies
Japan Patient Association
Malaysian Rare Disorders Society
MCT8-AHDS Foundation
Naevus Global
National Alliance for Rare Diseases Support – Malta
National Organisation for Rare Diseases of Serbia
National Organization for Rare Disorders
New Zealand Organisation for Rare Disorders
Nordic hypoPARA Organisation
Organization for Rare Diseases India
Pan Cyprian Federation of Patients’ Associations and Friends
Philippine Society for Orphan Disorders
Pro Rare Austria
Pulmonary Hypertension Latin Society
Rare Disease Ghana Initiative
Rare Disease Foundation of Iran
Rare Diseases South Africa
Rare Diseases Sweden
Rare Voices Australia
Retina International
Romanian National Alliance for Rare Diseases
Russian Patient Association
Thalassemia International Federation
The Ehlers-Danlos Society
The Federation of Esophageal Atresia and Tracheo-Esophageal Fistula Support Groups
World Alliance of Pituitary Organizations
World Federation of Hemophilia