Rare Diseases International (RDI) is the global alliance of rare disease patient organizations across all countries and across all rare diseases.

RDI’s **VISION** is a world where Persons Living with a Rare Disease (PLWRD) and their families experience a better life through full recognition and support.

RDI’s **MISSION** is to be a strong common voice for PLWRD and their families around the world.

The objectives of RDI are to:

- **ADVOCATE** for rare diseases as an international policy priority;
- **REPRESENT** PLWRD and their families at international institutions;
- **SUPPORT** the empowerment of RDI Members to drive local, national and regional change.

RDI’s Members include national alliances of rare disease patient organizations, regional organizations, and disease-specific international federations that altogether covered over 150 countries by the end of 2021.

RDI’s strategy is defined by RDI Members and RDI governance structures (RDI Council and the Board of Officers).

**Milestones in 2021**

**1. Governance**

RDI was incorporated under French law as an “Association Loi 1901” on 9 October 2018 and has been operating as a legally registered organization since January 2019. Overall, **RDI has 8 years of existence**: 3 years of existence with its own legal identity 2019-2021 and 5 years as a self-governed organization supported by a programme of one of its founding members, EURORDIS–Rare Diseases Europe.

The Statutes (constitution) and the By-Laws (internal rules) of the organization are available on the website: [https://www.rarediseasesinternational.org/rdi-council-governance/](https://www.rarediseasesinternational.org/rdi-council-governance/)
The RDI Council unanimously adopted revised By-Laws on 09 April 2020. These By-Laws are the internal rules for good governance, complementing the RDI’s Statutes. The revised By-Laws were presented and adopted at the General Assembly in May 2020, and no change has been made in 2021.

2. RDI Council Elections

The RDI Council is elected by Full Members and ensures effective governance of RDI. It also advises on RDI’s long-term strategy and actions.

Council elections were organized in April 2021, and all Full Members of RDI had the opportunity to vote online to replace or re-elect the following outgoing Members of the Council:

- **Lisa Sarfaty**, National Organization for Rare Disorders (NORD), term 2018 – 2021;
- **Rachel Yang**, Chinese Organization for Rare Disorders (CHARD), term 2018 – 2021;

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<thead>
<tr>
<th>Council Member</th>
<th>Organization</th>
<th>Year of Election</th>
<th>End of Mandate</th>
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<tr>
<td>Lisa Sarfaty</td>
<td>National Organization for Rare Disorders (NORD)</td>
<td>2018</td>
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<td>Rachel Yang</td>
<td>Chinese Organization for Rare Disorders (CHARD)</td>
<td>2018</td>
<td>2021</td>
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<td>Ritu Jain</td>
<td>Dystrophic Epidermolysis Bullosa Research Association (DEBRA) International</td>
<td>2018</td>
<td>2021</td>
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<td>Name</td>
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<tr>
<td>Durhane Wong-Rieger</td>
<td>Canadian Organization for Rare Disorders (CORD)</td>
<td>2019</td>
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<tr>
<td>Yann Le Cam</td>
<td>EURORDIS - Rare Diseases Europe</td>
<td>2019</td>
<td>2022</td>
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<tr>
<td>Alba Ancochea</td>
<td>Federación Española de Enfermedades Raras (FEDER)</td>
<td>2020</td>
<td>2023</td>
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<tr>
<td>Kin Ping Tsang</td>
<td>Rare Disease Hong Kong (RDHK)</td>
<td>2020</td>
<td>2023</td>
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A total of 12 nominations were received, and 39 Full Members voted. Kelly du Plessis, nominated by Rare Diseases South Africa (RDSA), was elected with 15 votes. Rachel Yang, nominated by both China Alliance for Rare Diseases (CHARD) and Illness Challenge Foundation (ICF) was re-elected with 17 votes. Ritu Jain, nominated by DEBRA International, was re-elected with 17 votes.

RDI Council 2021 AFTER the Council elections in April 2021
3. RDI Secretariat

STAFF

In 2021, four staff members joined RDI, two permanent and two temporary staff members:

Ayda Ramazzina joined RDI in January 2021 as the Governance and Development Manager. Ayda ensures that RDI remains a well-governed and sustainable organization. She works closely with the RDI Council, leads RDI’s resource development activities, and is the main contact for RDI’s donors.

Mary Wang joined RDI in September 2021 as the Science Policy Manager. Mary develops and coordinates RDI’s initiatives related to access to diagnosis and treatment. Specifically, Mary manages the development of an Operational Description of rare diseases, and the analysis of WHO Model Lists of Essential Medicines and Essential In Vitro Diagnostics. Both activities are developed under the MoU between the WHO and RDI. She also supports the coordination of the IRDiRC-RDI Global Access Working Group to initiate research into barriers to accessing rare disease medicines.

Nida Hasan joined RDI in September 2021, after her internship period, as the Global Capacity Building Junior Manager. Nida supports RDI’s international advocacy activities, the work on the Collaborative Global Network for Rare Diseases, and the RDI Capacity Building Programme.

Laura Philidor joined RDI in December 2021 as the Communications Junior Manager. Laura supports RDI’s communication specific to the core advocacy activities and the establishment of the global communications strategy.

In December 2021, Clara Hervas – RDI Public Affairs Manager from 2018 to 2021 – moved to another position in Brussels within the private sector. The recruitment to replace Clara took place at the end of 2021.

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RDI would like to warmly thank Clara for her work and dedication to RDI, and her invaluable contribution to RDI’s advocacy work, especially on the integration of rare diseases in the Political Declaration on UHC and the UN Resolution on addressing the challenges of PLWRD and their families.

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INTERNSHIPS

Nida Hasan worked at RDI as an Intern from May to August 2021, supporting the international advocacy activities and the work on the CGN4RD.

Emma Safar worked at RDI as an Inter from May to August 2021, supporting international advocacy activities.
VOLUNTEERS

In 2021, RDI also counted on the generosity of high-profile volunteers:

In New York, **Rasha Alnaibari** (until Summer 2021) and **Claudia Hirawat**, both are dedicated to the advocacy work towards the UN Resolution on addressing the challenges of PLWRD and their families. RDI warmly thanks Claudia for her continued personal commitment and time dedicated to engaging with the UN system at the highest professional quality standard.

In Fall 2021, **Noel Wathion** accepted to become the EURORDIS Representative for regulatory science and international policy as a Volunteer, thereby supporting the work of RDI specifically on access-related issues.

TEAM CHART (2021)

As the team grew in 2021, RDI held its first Secretariat “Strategic Planning” Workshop. This was a two-day offsite meeting with the following objectives:

- Ensure alignment between the **Action Plan and the Strategic approach**;

- Re-assess work priorities within the Team and ensure the **feasibility of the Action Plan**;

- Come together **face-to-face** as a new enlarged Team after the COVID-19 pandemic;

- During the meeting, the Team mapped all ongoing and expected activities across RDI’s priorities, identified areas of opportunities for improvement, and brainstormed a strategic narrative that encompasses all RDI’s activities. The outputs from this Workshop were:
  - Slides summarizing RDI’s strategic approach to creating local impact: enabling international environments while supporting national efforts to reinforce local impact;
  - A detailed Activity Tracker of RDI’s initiatives to fulfil the Action Plan including assigned Team lead and timeline;
  - A list of tasks and activities to undertake in the medium/long term;
  - Identification of RDI’s evolving human resources needs.
4. RDI Membership

In 2021, RDI’s membership continued to be engaged in RDI’s mission through representation in committees, working groups, and expert panels on topics that ranged from health care systems strengthening to awareness-raising and advocacy. Its growing and active membership increasingly reinforce RDI’s legitimacy and strength as the international rare disease movement.

Starting with 20 Member Organizations in 2015, By the end of 2021, RDI’s members included 81 national and regional organizations as well as disease-specific federations, gaining six new Members in the past year.

New Members include national alliances from the Ivory Coast and Georgia as well as four international federations for specific disease areas. Through its membership, RDI is active in 150 countries worldwide and on 6 continents.

The full list of RDI Members is regularly updated and published online at the following address: https://www.rarediseasesinternational.org/members-list/

Hlawulani Mkhabela, RDI Strategic Manager, manages and coordinates the membership programme.

Kin Ping Tsang, acting on behalf of the RDI Council, supports the review of new membership applications and makes recommendations regarding membership.

5. Communication & Engagement

In 2021, RDI further developed its communication message and visual identity as the voice of PLWRD and their families.

Through its communication channels, RDI aims to elevate the experiences of PLWRD and their families and showcase its diverse global membership. RDI is progressively integrating different elements in its communication strategy, keeping a formal and institutional approach when needed while highlighting its core identity as a grassroots alliance active at the global level.
In 2021, RDI launched a communication strategy with four key elements:

- Spotlighting the stories and images of PLWRD and families worldwide;
- Showcasing RDI Members;
- Privileging inclusive and accessible language (progressively translating materials in multiple languages);
- Using a bright and vibrant colour pallet (echoing the Rare Disease Day campaign).

This new approach is reflected in the new RDI website, PowerPoint Slide templates, printable conference materials, resources, toolkits, and videos produced in 2021.

NEW RDI WEBSITE

On 15 December 2021, RDI launched a new website with a user-friendly layout, pages and resources in multiple languages, and bright visuals putting the spotlight on the community of PLWRD and their families.
Visitors can learn about RDI’s mission and find a complete list of staff, Council Members, committees and working groups. Information about governance and funding is transparent and easy to locate. RDI advocacy priorities and global programmes are presented with relevant additional resources and materials.

The homepage includes a dynamic carousel of important milestones that have shaped rare disease advocacy and events around the world hosted by both RDI Members and other stakeholders. The Twitter feed can also be accessed on the homepage, enabling all visitors to follow RDI on social media.

The Members’ Page has been improved to allow visitors to view all Members easily, find Member contact information and filter organizations active at the national, regional, and international levels, including federations for specific disease areas.

RDI’s “Meet the Patient video Series” has migrated to the new site. The series features interviews with Members in Australia, Argentina, Ghana, Ireland, India, Mexico, Singapore, the United Kingdom, the USA, and the Netherlands.

The new “Get Involved” tab outlines how all stakeholders can be involved in RDI’s mission, including donating directly on the website, becoming a member, or joining the RDI Alliance of Companies for patient-centred action (AoC).

Webpages are available in English, French, Portuguese, and Spanish. Toolkit materials on Universal Health Coverage and the UN Resolution are available in nine languages: Arabic, English, French, Hindi, Japanese, Malay, Mandarin, Portuguese, and Spanish.

THE JOURNEY OF LIVING WITH A RARE DISEASE

On 13 September 2021, RDI launched “A Journey through Life with a Rare Disease”, an infographic illustrating the impact of living with a rare disease on PLWRD and their families.
Initially developed to support advocacy toward the UN Resolution, the infographic has become a vital part of RDI’s new communication strategy that focuses on the complex challenges faced by PLWRD and their families. The graphic is a journey around the globe following RDI’s character from birth through life - focusing on key life milestones and the challenges associated with finding a school and accessing quality education, transitioning to adult life, finding decent work, and fully participating in society.

In October 2021, the infographic was brought to life as a short, animated video that features on the new RDI website: [https://youtu.be/medJngPkfEg](https://youtu.be/medJngPkfEg)

**SOCIAL MEDIA**

In 2021, RDI increased its visibility on social media and leveraged social channels to reinforce international advocacy priorities and reach out to the broader civil society community.
Over the year, the **Twitter** following expanded with 913 new organic followers. Highlights include the announcement of the adoption of the UN Resolution, which was shared by a record-breaking 140 Twitter accounts.

Over 500 new followers joined RDI **LinkedIn** in 2021. Through the platform, RDI disseminated advocacy statements, posted recruitment announcements, and followed thought leaders in the field of rare diseases.

The RDI **Facebook** Page was launched in November 2021 to extend RDI’s public visibility and connect with smaller rare disease organizations around the world.

**TOOLKITS AND RESOURCE**

Clear and memorable communication reinforced advocacy efforts and campaigns on RDI’s priorities. The Communication Team supported the development of tools and communication on Universal Health Coverage (UHC4RareDiseases) and the UN Resolution (Resolution4Rare).

#Resolution4Rare

Through consistent and regular communication, RDI has been keeping its network engaged throughout the year, as well as the broader rare disease community informed of developments in the advocacy campaign toward the adoption of the UN Resolution.

In 2021, a logo and brand were developed to support the call for a UN Resolution.
At each stage of the campaign, tailored materials were created. In the first few months of the campaign, videos and social media described the UN system and the value of a UN Resolution. As the campaign evolved, RDI produced a series of materials in English, Spanish, Portuguese, and French to highlight the link between the Sustainable Development Goals (SDGs) and the challenges of PLWRD. The goal was to empower the whole community to advocate for the UN Resolution within the UN Agenda 2030, its Sustainable Development Goals, and the commitment to “Leave No-one Behind”.

The Resolution4Rare campaign received significant attention on social media, and posts related to the Resolution were broadly relayed and shared by RDI Members.

In 2021, the UHC4RareDiseases factsheet was translated into eight additional languages: Arabic, French, Hindi, Japanese, Malay, Mandarin, Portuguese, and Spanish. The factsheet is designed to empower patient advocates to speak to their national policymakers and promote UHC policies in their country for Persons Living with a Rare Disease.

The monthly electronic newsletter provides an overview of important updates, news, and events about RDI and the international rare disease community. The RDI Newsletter is published every month, serves as a rundown of key developments, and highlights opportunities for collaboration and engagement in RDI activities. The newsletter is also available on the RDI website.
Media Highlights

- **Leave no one behind – UN resolution for families living with a rare disease**, Health Awareness, 2021
- **Rare diseases are often ignored by public policies, the journey towards full inclusion is long**, Non-profit, 2021
- **Making rare diseases a human rights priority**, Health Awareness, 2021
- **Qatar affirms commitment to actively contribute to international efforts to achieve comprehensive, sustainable development**, Gulf Times, 2021
- **Letizia takes the floor before the United Nations on Rare Diseases**, Market Research Telecast, 2021

Hlawulani Mkhabela leads the Communications & Engagement Team. Hlawulani oversees the content and design of the RDI website with technical assistance from Davor Duboka, EURORDIS Web Technology Manager.

Laura Philidor joined RDI as the Junior Communications Manager in December 2021. Laura manages RDI Social Media and supports engagement and outreach initiatives.

**Rare Disease Day**

Rare Disease Day (RDD) is the international awareness-raising campaign coordinated by EURORDIS and led by over 60 national alliances worldwide.

Over the last years, the campaign has called for equity in social opportunity, healthcare, and access to diagnosis and therapies for PLWRD.

RDI ensures that a diverse community of patient advocate groups continues to shape the campaign. In 2021, RDI joined the RDD Steering Committee, a consultative committee defining RDD’s strategic objectives and priorities.

RDI member national alliances are official partners of the Rare Disease Day campaign and are included in the RDD Outreach and Communication Working Group. National alliances for Kenya, Zimbabwe and Georgia joined the list of RDD official partners in 2021. RDI collaborates with RDD to amplify advocacy messages and launch key campaigns. In 2021, RDI together with civil society partners EURORDIS and the NGO Committee for Rare Diseases launched the call for the UN Resolution on 4 March as a way to mark RDD 2021.

The RDD campaign disseminated branded materials on the UN Resolution using its social media platforms, with over 100 000 followers on Twitter, Instagram, and Facebook.
RDI also contributed to updating the equity factsheet included in the 2021 RDD toolkit. The factsheet places rare disease advocacy within the social equity and human rights movement and outlines milestones in international advocacy.

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<tr>
<th>RDI Rare DiseaseDay Partners 2021</th>
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<tr>
<td>Arabic Organisation for Rare Diseases</td>
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<tr>
<td>Associação Brasileira de Enfermidades Raras</td>
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<td>Botswana Organisation for Rare Diseases</td>
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<td>Canadian Organization for Rare Disorders</td>
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<td>Instituto Vidas Raras</td>
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<td>Chinese Organization for Rare Disorders</td>
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<td>Federación Colombiana de Enfermedades Raras</td>
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<td>Rare Disorders Kenya</td>
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6. RDI 7th Annual Meetings

RDI GENERAL ASSEMBLY

The 7th General Assembly took place on 25 May 2021. The meeting was held completely online in response to restrictions imposed by the COVID-19 pandemic.

67 patient representatives from RDI Member Organizations took part in the General Assembly. Before the General Assembly, RDI Full Members voted online on Resolutions to adopt the following documents:

- Activity Report 2020;
- Financial Reports for the years 2019 and 2020;
- Auditor’s Reports for the years 2019 and 2020;
- Auditor’s Special Report on regulated conventions for years 2019 and 2020;
- Action Plan 2021 and Budget 2021;
- Membership fees 2021.

RDI MEMBERSHIP MEETING

The 7th Membership Meeting was hosted on 26 May 2021 and attended by 82 participants from Member Organizations in over 30 countries.

The RDI Membership Meeting is an annual gathering that offers all members an opportunity to shape RDI actions and messages. The 2021 Meeting focused on: the UN Resolution campaign, the Global Network for Rare Diseases, and the development of initiatives promoting global access to medicines.

At the Membership Meeting, the “Dear UN...” initiative was launched – a request for testimonies from the community to support the call for a UN Resolution. Over 100 contributions were made online. The stories and images collected became a central part of the communication and outreach strategy.


The agenda included presentations from the Federación Argentina de Enfermedades Poco Frecuentes, Malaysian Rare Disorders Society, Rare Disease Ghana Initiative, National Organization for Rare Disorders (NORD, USA), World Duchenne Organisation and Orphanet.
7. Building a community of global patient advocate

RDI’S ADVOCACY COMMITTEE

The Advocacy Committee is responsible for developing and recommending priorities, initiatives and policies for RDI, preparing position papers, statements or declarations, as well as reviewing any position, statement, declaration or petition considered for endorsement by the RDI Council.

Yann Le Cam, RDI Council Member and Chief Executive Officer of EURORDIS, is the Chair of the Advocacy Committee. Members of this Committee are bringing their knowledge and advocacy expertise to RDI. They are all patient representatives from 17 of RDI’s Member Organizations, and one representative from the International Alliance of Patient Organizations (IAPO), took part in this Committee in 2021 and represents a wide range of countries and diseases.

Christine White, from CORD (Canada), joined the Advocacy Committee in 2021.

Clara Hervas, RDI’s Public Affairs Manager, coordinated the work of the RDI Advocacy Committee throughout 2021, engaging its Members in a process of co-creation of advocacy campaigns via meetings and internal consultations, as well as keeping them informed on advocacy files of relevance to RDI.

In 2021, the RDI’s Advocacy Committee contributed to the following activities:

- Main and foremost: the Draft UN Resolution on addressing the challenges of PLWRD and their families, discussing the main asks and revising the contentious points;

- Preparation of the UHC Day Event “Strengthening Care Systems to achieve Equity”;

[Image of Committee Members]
As the COVID-19 pandemic continued to aggravate the health, social and economic inequalities experienced by PLWRD and their families, access to information and vaccines is essential. In response to the ongoing pandemic, RDI disseminated reliable information resources and joined global initiatives calling for equitable access to COVID-19 vaccines.

In 2021, the COVID-19 Resource Centre, a dedicated page on the website, was regularly updated. The webpage offers information resources created by regional and national rare disease civil society organizations. The platform collates links to articles, surveys, and infographics on the impact of the pandemic on the community, as well as statements and press releases from RDI Members, and care guidelines developed by disease-specific federations.

In March 2021, RDI signed the "WHO Declaration on Vaccine Equity", calling for greater access to COVID-19 vaccinations, diagnosis, and treatment.

From April 2021, RDI joined campaigns calling on world leaders and all relevant stakeholders to urgently take steps to ensure equitable access to COVID-19 vaccine.

In April, RDI also took part in the WHO's #VaccinEquity campaign by posting content on social media to raise awareness of the issue and its impact on vulnerable groups, including PLWRD.

On 3 November, RDI hosted a group discussion on access to COVID19 vaccines in low-and-middle-income countries, led by Francois Houyiez, EURORDIS Access to Therapies Director. Participants joined from France, Guinea, Italy, Peru, Sweden, the USA, and Zimbabwe to share their concerns about the distribution of vaccines and learn more about the international initiatives to improve access.

COVID-19 RESPONSE
8. Partnering with other sectors to build a sustainable organization

RDI ALLIANCE OF COMPANIES FOR PATIENT-CENTERED ACTION

The RDI Alliance of Companies for Patient-Centered Action (AoC) provides a platform for companies to support the rare disease cause internationally. The AoC is RDI’s primary means of engaging with companies active in the rare disease field. Companies that join the AoC have the opportunity to increase their understanding of the international rare disease community and landscape, to inform and contribute to its success.

The RDI Alliance of Companies aims to:

- Build an international network of companies active in rare diseases;
- Increase mutual understanding of issues of common interest;
- Create a unique platform for dialogue through:
  - Alliance of Companies Meetings: 2 to 4 meetings per year for companies to provide input on specific projects and for RDI to share updates on its activities;
  - Regular RDI events for all stakeholders;
- Stimulate companies to address the needs of PLWRD.

In 2021, the following 12 companies joined the AoC:

![Company Logos](image)

The RDI Secretariat organized three online meetings with the members of the AoC in 2021, more specifically in May, June, and December:

- On 21 May 2021, RDI presented the Collaborative Global Network for Rare Diseases to share progress on the programme and obtain perspectives from companies;
- On 8 June 2021, RDI introduced the Global State of the Art Resource to present the Resource in-depth and discuss activities and areas of common interest. Moreover, RDI provided updates on the campaign towards a UN Resolution on Addressing the Challenges of Persons Living With a Rare Disease and their Families, focusing on the five key asks from civil society;
Finally, the meeting on 1 December 2021 focused on **RDI’s positioning in the global access discussion**, provided an opportunity to present RDI’s activities, and discuss access in Low-and Middle-Income Countries. RDI also shared the latest information and progresses towards the adoption of the UN Resolution on addressing the challenges of PLWRD and their families.
International Advocacy Activities

CALL FOR A UNITED NATIONS GENERAL ASSEMBLY (UNGA) RESOLUTION ON ADDRESSING THE CHALLENGES OF PLWRD AND THEIR FAMILIES

As of January 2021, RDI together with EURORDIS and the NGO Committee for Rare Diseases launched an intense advocacy process in two parallel ways: a grassroots campaign #Resolution4Rare reaching out to and mobilizing RDI’s and EURORDIS’ members to act nationally; and a policy campaign targeting Permanent Representations of the UN Member States in New York while working closely with the Core Group of Member States (Spain, Brazil, and Qatar) – the group of countries promoting the Resolution within the UN.

RDI also led the development of the campaign’s visual identity and Social Media strategy which initially served to inform patient organizations about the UN system and the relevance of international texts and was later crucial to amplifying the call for a UN resolution in the public and policy space.

While the campaign was steadily progressing, the process was marked by several key moments during 2021, as follows:

- **On 4 March 2021**, the NGO Committee for Rare Diseases held its Third High-Level Event, organized online by RDI and EURORDIS, to mark Rare Disease Day 2021 and launch the global call for the United Nations General Assembly to adopt, at its 76th Session, a Resolution addressing the challenges of PLWRD and their families.

Civil society partners described their five Key Asks for the Resolution and presented a video of support to the cause of PLWRD at the UN. The High-Level Event included contributions from UN Member States representatives from Brazil, Qatar, Spain, France, Thailand, and Japan.
In addition, Nata Menabde, Executive Director at the WHO Office at the UN, and Todd Howland, Chief of the Development and Economic and Social Issues Branch at the OHCHR, issued a statement of support.

Watch the Event: https://www.youtube.com/watch?v=oMbO0tyRFyc

In addition, Nata Menabde, Executive Director at the WHO Office at the UN, and Todd Howland, Chief of the Development and Economic and Social Issues Branch at the OHCHR, issued a statement of support.

On 7 July 2021: Official Side-Event to the UN High-Level Political Forum (HLPF) on Sustainable Development co-sponsored by the Core Group of Member States tabling the UN Resolution on PLWRD.

The event was entitled “On the road towards COVID-19 recovery and delivery of the SDGs: Addressing the challenges of persons living with a rare disease as a sustainable development, human rights, and equity priority”. It was officially co-sponsored by the Permanent Missions to the UN of Spain, Brazil, and the State of Qatar, together with RDI, the NGO Committee for Rare Diseases, and EURORDIS.

The online event was attended by over 300 participants from 61 countries across the globe and was opened by high-level representatives from the Core Group with a keynote address by Mr Craig Mokhiber, Director of the New York Office of the OHCHR. Discussions revolved around how addressing the challenges faced by PLWRD advances the UN 2030 Agenda, its Sustainable Development Goals, and the commitment to “Leave No One Behind”.

Watch the Event Highlights: https://www.youtube.com/embed/ajZgWTKI7l0

• On 7 July 2021: Official Side-Event to the UN High-Level Political Forum (HLPF) on Sustainable Development co-sponsored by the Core Group of Member States tabling the UN Resolution on PLWRD.

• I7 – 9 September 2021: UN Regional Group Meetings on the UN Resolution

RDI, together with EURORDIS, NGO Committee for Rare Diseases and Core Group of Member States co-organised five UN Regional Group meetings as well as one specifically targeting the EU Delegation to the UN. These closed meetings took place between 7 and 9 September and were the opportunity for additional Member States to hear from civil society the testimonies on the value of a UN Resolution directly from PLWRD and their families.

It was also the opportunity for the Core Group to provide details on the informal negotiations that they were launching ahead of discussions at the Third Committee of the UNGA. These informal negotiations, planned for the end of September and the beginning of October aimed at engaging positively with the Member States and reaching a consensus on the UN Resolution.
October and November 2021: time for negotiations and adoption by consensus by the Third Committee of the UNGA

Intense official and unofficial negotiations were led and organized by the Core Group of Member States promoting the UN Resolution within the UN towards the Permanent Representations in New York. During this pivotal two-month period, several key issues were raised by different UN Member States that could endanger the content of the UN Resolution on addressing the challenges of PLWRD and their families. The Core Group of Member States constantly collaborated with RDI to respond to concerns raised on different issues until the adoption by consensus of the Resolution by the Third Committee of the UNGA on 15 November.

During this period, RDI attended the World Orphan Drug Congress and was able to update attendees in person and online on the adoption by the Third Committee, making sure that civil society’s key messages and asks were appropriately communicated to a broad range of stakeholders, also starting to pave the way for the next steps in the coming years.

On 16 December 2021, the UN Resolution on “Addressing the challenges of PLWRD and their families” was formally adopted by consensus by the UN General Assembly.

There is an estimated 300 million persons living with a rare disease worldwide (PLWRD)

There are over 6,000 different rare diseases, most of which start in childhood.

Rare diseases can be genetic, rare cancers, rare infections and allergies (bacterial, viral, or caused by factors like food poisoning or chemicals).

Rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening.

Knowledge and information is scarce and persons living with a rare disease cannot access expertise.

The lack of awareness and recognition leads to discrimination and specific challenges in healthcare, education, employment and leisure.

As a result, persons living with a rare disease are psychologically, socially, culturally and economically vulnerable populations.

A UN General Assembly resolution should call for:

1. Human rights and inclusion: Participation and inclusion of persons living with a rare disease and their families in society and respect of their human rights.

2. Appropriate care: Improvement of health and social outcomes with the appropriate care and support within existing resources.


4. Recognition in the UN system: Integration and visibility of the rare diseases issue into UN agencies and programmes.

5. Monitor progress and implementation: Regular reports by the UN Secretariat to monitor the implementation and progress on the status of persons living with a rare disease.
As previously mentioned in the context of the adoption of the UN Resolution, on 4 March 2021, RDI hosted a policy event to mark Rare Disease Day, in partnership with the NGO Committee for Rare Diseases and EURORDIS. The event, entitled “Call for a UN Resolution on Persons Living with a Rare Disease – Why a UN Resolution & Why Now”, launched the international campaign for a UNGA Resolution.

261 participants from 83 countries attended the event, which brought together civil society organizations, PLWRD and families, UN Representatives, UN Member States, and policymakers.

The overarching goal of the event was to firmly position PLWRD as a vulnerable population requiring international action within the UN Agenda2030. Indeed, to achieve the Sustainable Development Goals and “Leave No One Behind”, it is necessary to address the needs and challenges of PLWRD.

At this event, civil society partners presented the Five Key Asks from the rare disease community as expressed in the Draft UN Resolution.

PLWRD and their families, represented by patient advocates from New Zealand, Brazil, Malta, and Kenya, appealed to the world’s leaders, describing what it means to live with a rare disease and highlighting the potential impact of a UN Resolution.

Amongst other participants, Nata Menabde, Executive Director at World Health Organization, affirmed that health systems are not adapted for rare diseases and the needs of PLWRD. Todd Howland, Chief of Branch on the Right to Development, Sustainable Development, and Economic and Social Rights Branch at the Office of the High Commissioner for Human Rights underlined that the lack of awareness and visibility fostered discrimination and exclusion of PLWRD and their families.

This was also the first opportunity for the Member States to show support for the proposed Resolution.

Messages of support were shared by the following UN Member State Representatives:

- H.E. Ambassador Sheikha Alya Ahmed bin Saif Al-Thani, Permanent Representative of the State of Qatar to the United Nations;

- H.E. Ambassador Agustin Santos Maraver, Permanent Representative of Spain to the United Nations;
• H.E. Ambassador Ronaldo Costa Filho, Permanent Representative of Brazil to the United Nations;

• H.E. Ambassador Nicolas de Rivière, Permanent Representative of France to the United Nations;

• H.E Ambassador Vitavas Srivihok, Permanent Representative of the Kingdom of Thailand to the United Nations;


Universal Health Coverage (UHC) ensures all people have equitable access to diagnosis, treatment, and care without facing financial hardship. For UHC to indeed be universal and cover all vulnerable communities, UHC policies must address the needs of PLWRD.

In September 2019, RDI successfully advocated for the inclusion of rare diseases in the UN Political Declaration on Universal Health Coverage. Since then, RDI has been developing specific toolkits and advocacy materials that are publicly available to support rare disease patient organizations, and in particular national alliances, to advance UHC national policies and measures. Indeed, after securing a global framework, it is necessary to support national efforts to advocate for UHC for rare diseases at the national level, towards national authorities and policymakers to make a real difference for PLWRD and their families.

#UHC4RareDiseases - Universal Health Coverage for Rare Diseases Campaign

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UHC2030

RDI is a partner of UHC2030 and a member of the UHC2030 CSEM, the Civil Society Engagement Mechanism, a body founded by the World Bank and the World Health Organization to advance equitable UHC policies and action. As such, RDI is committed to driving support for UHC and promoting the delivery of high-quality health and social care to all PLWRD around the world.

In September 2021, the CSEM developed the Health for All Advocacy Toolkit to strengthen capacity and mobilize civil society in support of UHC. The #UHC4Rare Toolkit of RDI appears as an essential resource in the CSEM’s toolkit, reflecting the growing visibility of the rare disease community in the international UHC movement.
In December, RDI traditionally holds every year a Global Policy Event on **UHC Day**, which is officially on 12 December, to promote Universal Health Coverage for rare diseases.

This year, RDI partnered with the CSEM to host, on Monday 13 December, an in-depth discussion on health equity for PLWRD, entitled: "Strengthening Care Systems to Achieve Equity."

The aim of this event was to send a strong message to governments and policymakers that to achieve Universal Health Coverage and "Leave No One Behind" by 2030, the focus needs to be on Equity. For the 300 million PLWRD worldwide, this means understanding the systemic reasons why they face barriers to good quality and affordable health care and then incorporating targeted policy interventions to address those barriers within person-centred care systems. It also means pursuing health-in-all-policies and interventions that encompass the entire life course, as well as the interrelated aspects of physical, mental, and social health and wellbeing.

At the event, **RDI released its Policy Statement** entitled "Health Equity: The perspective of Persons Living with a Rare Disease". The paper was presented at the UHC event and will serve as an advocacy tool for RDI and its members.

The event opened with statements from Vytenis Andriukaitis, WHO Special Envoy for UHC and Gabriela Cuevas Baron, Co-chair of UHC2030 Steering. The rare disease community was represented by Rare Diseases South Africa, Federación Colombiana de Enfermedades Raras and DEBRA International.

**#UHC4Rare Toolkit**

The **#UHC4Rare Toolkit** provides practical tools for patient advocates to raise awareness and ask policymakers to safeguard equity and consider the needs of PLWRD in national UHC strategies.

In 2021, RDI updated the **Universal Health Coverage for Rare Diseases Toolkit** #UHC4RareDiseases by translating the campaign factsheet into eight additional languages: Arabic, French, Hindi, Japanese, Malay, Mandarin, Portuguese, and Spanish.

**NONCOMMUNICABLE DISEASES (NCDS)**

The NCD Alliance hosted a round table discussion entitled "Leaving No-one Behind: The Need for a Person-Centered, Inclusive NCD Agenda", on 5 November 2021. RDI Chair, Durhane Wong-Rieger participated in the panel discussion, offering a perspective from the rare disease community.
Global Initiatives and Programmes: Supporting access to diagnostics, treatments, and care

1. Memorandum of Understanding between WHO and RDI: Second year of execution

OPERATIONAL DESCRIPTION FRAMEWORK FOR RARE DISEASES

The **first deliverable describing the conceptual framework to develop an operational description of rare diseases was submitted to the WHO in March 2021** (WHO D0). Subsequently, a multi-stakeholder group of **18 experts from six continents** were invited by RDI to participate in the development of the operational description of rare diseases in detail. The participants included clinicians and researchers (e.g., paediatricians, geneticists), terminology producers (i.e., for health information systems), medical statisticians, industry experts, patient advocates, policymakers, and public health experts. Two Medical Officers from the Classifications and Terminologies (ICD, ICF, ICHI), Department of Health Statistics and Information Systems of WHO participated and contributed to the expert group.

<table>
<thead>
<tr>
<th>Expert Panel</th>
<th>Iberoamerican Alliance for Rare Diseases (ALIBER), Federación Argentina de Enfermedades Poco Frecuentes (FADEPOF), Argentina</th>
<th>Jane Millar</th>
<th>International Health Terminology Standards Development Organisation (IHTSDO), SNOMED International, UK</th>
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<tbody>
<tr>
<td>Roberta Anido de Pena</td>
<td>Chiesi Pharmaceuticals, Italy</td>
<td>Caron Molster</td>
<td>Office of Population Health Genomics, West Australia Department of Health, Australia</td>
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<tr>
<td>Diego Ardigò</td>
<td>Rare Care- Clinical Centre of Expertise for Rare and Undiagnosed Diseases, Perth Children’s Hospital, Western Australia, Australia</td>
<td>Lucia Monaco</td>
<td>International Rare Diseases Research Consortium, Fondazione Telethon, Italy</td>
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<tr>
<td>Gareth Baynam</td>
<td>DA Precision Health, Australia</td>
<td>Carmencita Padilla</td>
<td>University of the Philippines Manila, Philippines</td>
</tr>
<tr>
<td>Hugh Dawkins</td>
<td>Johns Hopkins School of Medicine, Online Mendelian Inheritance in Man (OMIM), USA</td>
<td>Anne Pariser</td>
<td>National Center for Advancing Translational Sciences (NCATS), NIH, USA</td>
</tr>
<tr>
<td>Ada Hamosh</td>
<td>Health Statistics and Information Systems, World Health Organisation, Switzerland</td>
<td>Ana Rath</td>
<td>Orphanet, INSERM, France</td>
</tr>
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An online, modified Delphi approach was used to engage the expert group in developing and building a consensus on the operational description of rare diseases. The approach contains multiple rounds of review and prioritization via 3 online workshops and 2 surveys to enable structured discussion between November 2021 and January 2022.

For the first workshop, the experts provided statements on the essential elements for a definition of rare diseases. The key concepts were organized into a survey and scored by the experts before the second workshop. Survey statements addressed: Objectives and elements of an operational description with statements focused on terminology, the descriptor of ‘disease’, the quantitative descriptor of ‘rarity’, and qualitative descriptors. Considering the results of the two surveys together with discussions during the first and second workshops, a first outline of the operational description of rare diseases was developed at the end of 2021 for discussion during the final workshop in January 2022.

COLLABORATIVE GLOBAL NETWORK FOR RARE DISEASES

RDI has continued to lead a five-year Work Programme, supported by the Technical Research, to coordinate activities with the rare disease community to scope out and develop a global network for rare diseases, referred to as the Global Rare Diseases Network (GRDN) and previously named Collaborative Global Network for Rare Diseases (CGN).
The coordination of the activities continued to follow the planning documents - **Technical Research Plan** (bottom-up) and **Work Programme** (top-down), which had been previously endorsed by the WHO in 2020.

In the second year of the programme and MoU with the WHO, RDI has strengthened and deepened the collaboration with the World Health Organization to prepare for the launch of a pilot phase of the network. Based on the Need Assessment Study Report and the Concept Model developed in the first year of the MoU, RDI expanded the Concept Model into an initial draft for an Operational Framework that allows for a successful launch of the pilot phase of the GRDN. The research activities performed to support this reflection included:

- **a) Analysing** the operational governance structure, clustering proposal and sustainability models for the GRDN;
- **b) Co-designing** a prototype for the readiness appraisal;
- **c) Engaging** the rare disease community in a readiness appraisal for their participation in the pilot.

### Technical Research Progress:

The Technical Research in 2021-22 was conducted by RDI, in partnership with **Health Standards Organisation (HSO)** and with **an international Panel of Experts**. The Technical Research that was completed by Q1 2022 included the following activities:

1) **Expansion of the Concept Model into an Operational Framework** (Structure). Included the identification of existing collaborations and potential members; development of a rare disease clustering model; and reflections on sustainability models. Three Topic-Based Working Groups were established by volunteers from the Panel of Experts. Specifically:

- **a) Regional Working Group(s)** completed a mapping exercise of existing collaborations and networks and identified existing expert centres in each WHO region. A total of **54 members from 33 countries joined the working group(s)** for the following WHO regions and sub-regions: AFRO, AMRO, EMRO, EURO, LATAM, SEARO and WPRO. A series of **online workshops (x6)** was organised with representatives from the Panel of Experts to identify the existing collaborations, their respective regions, and sub-regions, and discuss the common cultures, languages and experiences that underpin these existing collaborations. A report was developed and presented to the Panel of Experts in October 2021, and the results were discussed and ratified. Two separate webinars were held to enable the participation of experts from different time zones. A **total of 120 experts attended the two webinars**;

- **b) Disease-Cluster Working Group reviewed** the different models for clustering through the lens of people with lived experience and provided recommendations on the optimal model for inclusive participation of stakeholders, clustering experts for collaboration, and organizing actions to address needs at a global level. 20 Experts from the Panel of Experts, from 13 countries collaborated to identify, map, and evaluate the most suitable disease cluster model to recommend for the pilot phase;
c) Innovative Financing Working Group reflected on the different innovative financing models that could be used to support the pilot network based on reviewing several case studies of existing partnerships, consortiums, and networks. A total of 14 members from 10 countries joined the working group. A series of online workshops took place in Q1 2022 to discuss the resourcing needs of the network to support sustainability and to identify relevant models based on needs.

2) Building a Readiness Appraisal Tool was co-designed and based on four guiding principles: health equity, person-centred care, shared accountability, and a quality improvement approach. The core set of technical criteria was developed to be adaptive and inclusive so that no community is left behind or excluded from the pilot communities, irrespective of their level of maturity and resources (Criteria).

The Readiness Appraisal Tool was based on the combined findings from and reflections on the following three evidence-informed research activities:

a) Reflection on the literature review findings of the models and methods of assessing services and networks (completed in 2021). Building on a literature review that identified the published evidence related to the assessment used to support the development of an assessment programme for European Reference Networks (ERNs), the team completed a review of the most recent published academic evidence published since 2015. The literature search yielded 1991 papers. A review and screening of the abstracts resulted in 157 papers that specifically address methods for assessing services or networks. Of these, after review of the full texts, 59 articles were included in the full data extraction and thematic review;

b) Projects review and reflections of lessons learned. The team has complimented the literature review with learnings gained from co-developing, testing and evaluating in cooperation with people with lived experience, organizational leaders, and system partner projects to identify and test the emerging domains for the readiness appraisal. Topics on health equity, quality improvement, and people-centred care have emerged. A prototype of the Readiness Appraisal Tool was co-designed together with the Panel of Experts. This prototype of the Readiness Appraisal Tool is comprised of five domains;

c) Validate that the Readiness Appraisal Tool contained the appropriate content presented in a user-friendly format; the team developed and launched a survey on the Readiness Appraisal Tool. The survey sought feedback on whether the intended audience felt they could use the Tool to stimulate discussions locally. Responses were received from each of the six WHO regions and contained both qualitative and quantitative data. This information will be used to update the final version of the Readiness Appraisal Tool. The Readiness Appraisal Tool was further developed into a Consultation Workbook, practical and user-friendly, that can be used by each regional collaboration or network to help them take the necessary actions to prepare for application to the Network Pilot as a regional hub. The Consultation Workbook was produced in four languages – Arabic, English, French and Spanish.

3) Preparations ahead of the Pilot (Readiness). The rare disease community was supported through a series of regional workshops, stimulating local dialogue, and promoting equitable engagement. A comprehensive analysis was completed of environmental forces, health competencies strengths, resources, and constraints, as well as entry barriers, competition, risks, incentives, and opportunities.
The Team adopted an equity-driven strategy for engagement to promote equity, diversity, and inclusion in the pilot phase, and leave no one behind. This means that while all communities are engaged with the same information, the approach can differ depending on the needs of local communities. For example, in areas that need more intensive support and engagement, the team will organize regional and sub-regional workshops e.g., a workshop to support the LATAM region is being organized with material and facilitation in Spanish.

A series of regional workshops (x8) took place in Q1 2022 using the Consultation Workbook to support the communities to create dialogue and facilitate meaningful conversations with two key purposes.

Firstly, to identify early adopters or innovators of the collaborative hubs, and secondly, to allow the community leaders and experts to determine whether they are ready to take part in the initial pilot phase of the network. This series of regional workshops are planned to continue throughout 2022 to support communities to be ready for the launch of the pilot phase.

The Technical Research for the Collaborative Global Network for Rare Diseases was supported by Pfizer, Roche, Sanofi-Genzyme, and Vertex in 2021.

Work Programme

An international Panel of Experts with lived experience and topical experts was established in Q1 2021. These experts volunteered their time and expertise to join topic-based working groups to conduct the Technical Research, and as a whole Panel, review and independently validate the research findings and inform the underpinning policy recommendations that formed the Operational Framework (WHO Deliverable). Since the Panel of Experts was formed, the number of experts has increased to over 230 experts, representing 101 countries (51.8% of the WHO Member States).

Throughout 2021, the Panel of Experts collaborated virtually, through a series of online workshops, feedback via email, and revision of documents on an online platform. The contribution of time from the Panel of Experts in 2020–21 is estimated at a total of 1000 hours.

A series of Panel of Expert workshops were held in November 2021 and February 2022 to present the supporting proposals that underpin the initial draft of the Operational Framework following the engagement of the Panel of Experts, the RDI team elaborated a draft proposal for the Operational Framework of the GRDN. The draft proposal was submitted to the WHO, the content and format of the framework were discussed, and based on the feedback received, the final version of the proposed Operational Framework, and its Annexes were submitted to the WHO on 30 March 2022.
WHO MODEL LIST OF ESSENTIAL MEDICINES

The WHO publishes and maintains the WHO Model List of Essential Medicines as a tool for countries to adopt for the development of their national essential medicines list in line with local priorities. WHO defines essential medicines as those that satisfy the priority healthcare needs of the population, which are intended to be available in functioning health systems at all times in adequate amounts, in appropriate dosage forms, with assured quality, and at prices individuals and the community can afford.

The third collaboration area identified within the RDI – WHO MoU is to promote access to health services via the facilitation of patient access to essential medicines. More specifically, the objective is to explore how a reference list of medicines and diagnostics for rare diseases could provide the Member States with a model from which to develop a national priority list and inform policies. To explore the concept of such a reference list for rare diseases, steps were taken in collaboration with the International Rare Diseases Research Consortium (IRDiRC) to map the list of medicines that are currently on the WHO Model Essential Medicines List (EML) and Essential Medicines List for Children (EMLc) that are used for rare diseases, and gap analysis to identify medicines and diagnostics that are essential for rare diseases and not included in the existing model lists. In 2021, the IRDiRC Rare Disease Treatment Access Working Group reviewed the collated draft list of essential medicines for rare diseases and published the paper “Essential List of Medicinal Products for Rare Diseases–Recommendations from the IRDiRC Rare Disease Treatment Access Working Group” in the Orphanet Journal of Rare Diseases. RDI Board of Officers Durhane Wong-Rieger and Rachel Yang were among the co-author of the article.

The IRDiRC List of Medicinal Products for Rare Diseases contains 204 medicines approved by the United States Food and Drug Administration (FDA), the United States Food and Drug Administration (FDA), the European Medicines Agency (EMA) and China’s National Medical Products Administration (NMPA). The list includes 125 chemical drugs and 79 biologics, including recombinant proteins, polyclonal/monoclonal antibodies, cell, and nucleic acid therapies. Of note, six drugs have indications for treating multiple rare diseases. The list is organized into seven disease categories: metabolic, neurologic, hematologic, anti-inflammatory, endocrine, pulmonary, immunologic, and miscellaneous category. A total of 134 diseases are covered, with the largest number of drugs listed in the metabolic diseases category.
The working list of medicinal products for rare diseases is available for consultation online: https://bit.ly/MedicineList4RDs

EURORDIS volunteer Noel Wathion supports the development of the RDI strategy related to the WHO Model List of Essential Medicines and the Model List of Essential In Vitro Diagnostics.

2. Global State of the Art Resources

In most countries, there is a lack of data and information on rare diseases with multiple consequences that are detrimental to Persons Living with a Rare Disease globally. In a constant strive to generate and share information, as well as improve dialogue between different stakeholders towards creating a global rare disease-friendly environment, in 2021, RDI began exploring further feasibility of a global resource of information on rare diseases. This “Resource” would be established by pooling together data in collaboration between patient organizations, clinicians, national authorities, and academia. To ensure optimal data quality, this centralization of data would be done progressively, in a stepwise approach.

In 2021, several activities took place to progress the work aimed at establishing the Global State of the Art Resource, planned to start in January 2023:

- Finalization of the Global State of the Art Concept Note and discussion within the RDI Council;
- Organization of RDI Members’ Webinar on the Global State of the Art Resource attended by 55 participants on 21 April;
- Presentation of the Global State of the Art Resource Project to the RDI Alliance of Companies on 6 June 2021;
- Global SoA Survey with the participation of 19 RDI Members, launched on the 5 May 2021; 3 areas of most interest were identified: National rare disease plans and strategies; International rare disease collaborations, research and care initiative; and Rare disease policy and activities at the UN and UN Agencies.
- The Project and Survey results were presented to the Membership Meeting on 26 May 2021;
The Agreement aimed at establishing a **Consortium between RDI, EURORDIS and Orphanet INSERM** has been drafted by lawyers and is being reviewed by partners since October 2021.
Supporting emerging patient groups and building capacities globally

1. RDI Capacity Building Programme

RDI’s Capacity Building Programme aims to support the empowerment of rare disease patient groups to:

- **Drive advocacy** at the local, national and regional levels
- **Engage meaningfully in global collaborations**
- **Shape RDI** programmes and strategic direction

Members were asked in 2020 to complete a comprehensive survey on their capacity building needs and international advocacy priorities. The results of this survey were then used in 2021 for the development of a medium-term capacity building strategy and activities for the period 2022-2025.

The proposed Capacity Building Programme, submitted to the RDI Council in December 2021, focuses on three levels of impact: RDI member organizations, individual advocate leaders, and the rare disease community at large. The programme is split into content streams based on Members’ interests and priorities, as well as on RDI activities, including Advocacy, Equity & Access, and Global Collaborations.

The programme offers multiple ways to engage, from facilitated access to existing community resources to member-led small group discussions, and fellowships to participate in community events.

As a result, the Conferences and Fellowship programme was strategically incorporated under the Capacity Building Programme. In 2021, it was decided that from 2022 onward, the Capacity Building Programme would be expanded to include other community engagement activities, such as the activities linked to the RDD campaign, to develop a more comprehensive “Strategic Engagement Programme”.

**Lisa Sarfaty**, former RDI Council Member and NORD Vice President of Community Engagement, has supported the development of RDI’s Strategic Engagement Programme to be launched in 2022.

More informations online: [https://www.rarediseasesinternational.org/rdi-capacity-building/](https://www.rarediseasesinternational.org/rdi-capacity-building/)
CONFERENCES AND FELLOWSHIPS IN 2021

Due to continuing travel restrictions adopted in response to the COVID-19 pandemic, most events supported by RDI in 2021 were hosted online. RDI supported the following events:

- **RE(ACT) Congress and IRDiRC Conference** 2021, 13–15 January (ticket fee waivers and media partnership). Ten RDI Members and patient advocates attended the event online;

- **World Orphan Drug Congress, USA**, 25–27 August 2021 (ticket fee waivers and media partnership).

  - Three RDI members received free places at the conference.
  - The following RDI speakers are featured on the agenda:
    - Flaminia Macchia – “Campaigning towards a UN Resolution on addressing the challenges of Persons Living with a Rare Disease and their families”;
    - Yann Le Cam – “Needs-led innovation for a future that leaves no rare disease patient behind”;

- **World Orphan Drug Congress, Europe**, 14–16 November 2021 (travel and accommodation fellowships and media partnership).

  - Four RDI members received travel and accommodation fellowship to attend the WODC Europe–China Alliance for Rare Disease, Georgian Foundation for Genetic and Rare Diseases, Federación Colombiana de Enfermedades Raras, and the Chinese Organization for Rare Disorders;
  - Flaminia Macchia described the advocacy campaign toward the UN Resolution and the Resolution’s adoption by consensus on 15 November 2021 by the Third Committee of the UNGA.

- **Asia Pacific Alliance of Rare Disease Organizations (APARDO) Annual Conference**, 14–15 December 2021, receiving a grant from RDI;

- **African Summit on Rare Diseases**, 1–3 December 2021, organized by RDGI in partnership with RDI. Please find more information below:

Rare Disease Ghana Initiative (RDGI), in partnership with RDI, hosted the **African Summit on Rare Diseases**, a hybrid two-day event in Accra, Ghana. 254 participants joined the event virtually and 84 participants joined the event in person in Accra, including PLWRD and their families, community leaders and national authorities.
The African Summit followed previous African rare disease conferences in Cape Town (2018) and Johannesburg (2019), led by Rare Diseases South Africa.

The African Summit in Accra aimed to showcase emerging multi-stakeholder collaborations led by the African civil society movement and promote a more equitable future for PLWRD and their families on the continent.

The agenda included speakers from 15 African countries and was attended by over 400 participants joining online and in person. Speakers included African civil society leaders, regional and national policymakers, the private sector and clinicians and researchers. Each addressed challenges and barriers to care, as well as innovative partnerships transforming advocacy, research, diagnosis, and access at the national, regional, and global levels.

The programme included a plenary with three sessions: an opening discussion on the national context for PLWRD in Ghana, a panel discussion on the regional state of play in Africa, and a review of global collaboration.

On the final day, RDI and RDGI facilitated three parallel thematic workshops:

- Care Management and Advocacy in Ghana (local focus);
- Strengthening an African Rare Disease Network (regional focus);
- Global and Regional Collaborations (regional and global focus).

Each workshop group developed five key recommendations and next steps, which were shared in the Conference Report and at the Global Policy Event “Rare Diseases – A Global Priority for Equity” (28 February 2022, EXPO Dubai).

The event also represented an opportunity to call for support for the UN Resolution in the African continent while supporting awareness-raising activities on rare diseases in Africa. The Summit paved the way to prepare for the necessary steps to be taken at the regional and national level towards the implementation of the key asks of the UN Resolution.

During the African Summit, the video on “A Journey through Life with a Rare Disease” screened for the first time with an African voice over.
MEMBERS-LED WEBINARS

“Knowledge-Sharing on Advanced Therapies”, March–April–June 2021, Canadian Organization for Rare Disorders (CORD)

A three-part online workshop series on Advanced Therapies offered an introduction to cell and gene therapies, case studies from the field of rare diseases, and frameworks to facilitate access. The webinars were hosted by CORD in partnership with RDI.

- Workshop 1: “Introduction to Gene Therapy, Gene Editing and Cell Therapy”, 8 March 2021;
- Workshop 2: “Gene Editing”, 14 April 2021;

2. RDI’s active partnership in the international rare disease ecosystem

THE NGO COMMITTEE FOR RARE DISEASES : RDI IS THE GLOBAL VOICE OF THE COMMITTEE

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), an important interface between the world’s NGOs and the UN system. The Committee is one in forty CoNGO Committees, approved by the CoNGO General Assembly in 2014 and instigated by the CoNGO President in 2015. The Committee was then publicly launched in November 2016 at its First High-Level Event at the United Nations.

The main objective of the NGO Committee for Rare Diseases is to bring visibility and understanding of rare diseases to the United Nations and align the issues of relevance to Persons Living With a Rare Disease with the UN 2030 Sustainable Development Agenda: the Sustainable Development Goals (SDGs). The current goal of the Committee is to work with its NGO partners toward the adoption of a UN General Assembly Resolution on Persons Living with a Rare Disease.

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 Ägrenska Centre for Rare Diseases. The Vice-Chair of the Committee is Yann Le Cam, CEO of EURORDIS-Rare Diseases Europe. The other Members of the Executive Board are the World Federation of Hemophilia, the International Federation for Spina Bifida and Hydrocephalus, the International Alliance of Patients’ Organizations, and the International Alliance of Women. RDI’s Chair, Durhane Wong-Rieger, is RDI’s representative with in the Executive Board of the NGO Committee for Rare Diseases.
INTERNATIONAL RARE DISEASE RESEARCH CONSORTIUM (IRDiRC):

RDI is a partner engaged in the governance and several actions

IRDiRC, the International Rare Disease Research Consortium, was officially launched in 2011, with the vision to enable all PLWRD to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention. IRDiRC experience in the past decade has shown that the international rare diseases research community is eager to share knowledge and experience and work collaboratively across borders to bring diagnoses and therapies to patients.

The revised set of goals for the decade 2017 – 2027 is to accelerate progress towards its ambitious vision.

The three specific goals are the following:

- **Goal 1**: All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently un-diagnosable individuals will enter a globally coordinated diagnostic and research pipeline;

- **Goal 2**: 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options;

- **Goal 3**: Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.

IRDiRC is governed through a Consortium Assembly, an Operating Committee, three Constituent Committees and three Scientific Committees, aided by ad hoc Task Forces. The Scientific Secretariat provides organizational and communications support and manages IRDiRC actions on strategic projects and activities.

In 2021, RDI took part and contributed to a series of meetings within the following IRDiRC structures:

- RDI is represented in the Consortium Assembly, IRDiRC’s main governing body. In 2021, there were five meetings of the Consortium Assembly: 21-22 January, 15 March, 19 May, 16 and 19 July, and 9-10 December. Key RDI activities regarding the launch of the UN Resolution campaign, consultation on CGN and member workshops on ATMP were reported in the May Roundtable Meeting. These activities were relevant for Goal 3 of IRDiRC.

- RDI is represented in the Patient Advocates Constituent Committee (PACC). The PACC is composed of 16 umbrella patient groups of which 14 are Member Organizations of RDI and RDI is itself represented by Durhane Wong-Rieger, as Chair of the PACC. Three PACC meetings took place in 2021 to identify specific challenges and develop a proposal for Task Forces. A proposal to set up a Task Force to recognize and analyze the impact on caregivers of PLWRD was submitted to the consortium assembly but it was not prioritized for activation.
RDI is represented, via Durhane Wong-Rieger being Chair of the PACC, in the Operating Committee. The Operating Committee meets monthly to prepare and advance IRDiRC activities, process information, and enable more effective management of the consortium as a whole.

In recognizing the urgent need from RDI members to improve access to treatment and care, in 2022, RDI proposed the creation of a Working Group (WG) within IRDiRC to tackle issues around access. The Rare Disease Treatment Access WG was launched in 2020, co-chaired by Durhane Wong-Rieger and Dr William A. Gahl. The objectives of the WG were to:

1) improve standards of care for persons living with rare diseases by promoting access to approved medicines;
2) initiate research into barriers to accessing rare disease medicines, especially in LMICs; and
3) define opportunities to address those barriers. To improve global access to rare disease medicines, the first step was undertaken to create a list of standard-of-care medicines. In 2021, after the final review of the collated lists of medicines approved by key regulatory agencies in the USA, European Union, and China for the treatment of rare conditions, a manuscript was developed for publication. Durhane Wong-Rieger and Rachel Yang are among the co-author of the paper “Essential List of Medicinal Products for Rare Diseases – Recommendations from the IRDiRC Rare Disease Treatment Access Working Group” published in the Orphanet Journal of Rare Diseases in July 2021.

Since October 2021, the IRDiRC Rare Disease Treatment Access Working Group is supported by Mary Wang, RDI Science Policy Manager, becoming the IRDiRC-RDI Global Access Working Group. The WG was reorganized, and new members were recruited to participate in the new phase of the WG. The kick-off meeting of the new WG took place on 15 November 2021 online.

Matt Bolz-Johnson, RDI Programme Director, represents RDI in the new Task Force launched in November 2021 on Primary Care. PLWRD typically present first, and often recurrently, to their primary care providers. Primary care is therefore central to all aspects of the disease journey including referral pathways, diagnosis and care. The objectives of the Primary Care Task Force are aligned with the ambition and goals of RDI’s advocacy activities with the WHO, to establish a collaborative global network for rare diseases.

Dissemination of Task Force Call for Experts to RDI members as well as the nomination of members to specific Task Forces to promote patient advocate presence and inclusion in global initiatives. This has, in particular, resulted in many persons from the RDI community participating in the Enabling and Enhancing Tele health for Rare Diseases Across the Globe Task Force.

In 2021, RDI, via Durhane Wong-Rieger, contributed to the preparation of the IRDiRC Conference & RE(ACT) Congress (held on 13–15 January 2021): Preparation of conference sessions, presentation, and co-chairing of the session on “access to diagnostics and drug for all” and “patients as drivers in drug development and clinical trials”.

39
The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease, created by EURORDIS, Microsoft, and Takeda in 2018, is a multi-disciplinary group of experts committed to shortening the time to diagnosis by building a road map to improve diagnostic pathways for children with a rare disease.

The three co-chairs of the Global Commission are Gregory Moore (Corporate Vice President, Microsoft Health), Wolfram Nothaft (Chief Medical Officer, Takeda), and Yann Le Cam (Chief Executive Officer, EURORDIS-Rare Diseases Europe).

After EURORDIS facilitated and supported greater participation and visibility of RDI in the Global Commission Secretariat and work streams, RDI contributed to the overall vision, mission and activities of the Global Commission.

The primary objective of RDI’s involvement in the Global Commission is to actively promote greater integration of the patient voices in countries outside Europe and the USA in the Global Commission’s activities, thus supporting a more international perspective.

RDI is directly represented and involved in the activities of two Working Groups:

1. **The Empowerment & Awareness Working Group** (Flaminia Macchia is the Secretariat Lead) - Broaden awareness about the need to reduce the time to diagnosis and empower key stakeholders to accelerate diagnosis. In February 2021, the Rare Navigator Pilot was launched in San Diego, USA. RDI underlined the need to shape and adjust the Rare Navigator tool before expanding it to other countries and regions at a global level. RDI participated in the campaign’s Working Group meetings before the launch of the pilot phase and stands ready to contribute further to a true “internationalization” of the tool.

2. **The Policy & Advocacy Working Group** (Simone Boselli from EURORDIS is the Secretariat Lead) - Advocate for local and global policy change to build an enabling environment for rare disease diagnosis.

In 2021, RDI was involved in the reflection on the Global Commission structure and governance, and the identification of priorities in the coming years, based on further expansion of the international scope of the Global Commission activities. As such, RDI participated in two workshops in June and July 2021.

RDI also actively participated in the All Members Meeting in May 2021 and contributed to the reflection on Health Equity.
RDI also closely worked with the Global Commission and the EURORDIS Rare Barometer Team to expand the geographical coverage of the Survey on Diagnosis in countries outside of Europe, starting with six countries for a pilot phase at international.

**RDI drives the global survey on access to diagnosis with the Global Commission**

Delayed diagnosis, lack of diagnosis or multiple misdiagnoses are common challenges for PLWRD, with devastating consequences for patients and their families.

In 2021, RDI began collaborating with the EURORDIS Barometer Programme and the Global Commission to gather meaningful data on experiences of diagnosis and obstacles along the journey for PLWRD and their families.

"The journey to diagnosis for people living with rare diseases" is a survey project designed and managed by the Rare Barometer Programme, EURORDIS’ signature survey programme generating data on patient perspectives with a particular focus on Europe. RDI joined the initiative to leverage the EURORDIS’ Rare Barometer Programme to reach PLWRD and families globally. With support from the Global Commission, RDI extended outreach and engagement to 6 pilot countries:

Argentina, Australia, Brazil, Malaysia, South Africa, and the United Arab Emirates. Malay and Arabic were added to the 23 European languages already used by the survey programme to ensure participation from all six countries.

Throughout June and July 2021, RDI connected with its members in the pilot countries. Representatives from each country participated in the qualitative research phase to inform the final questionnaire and will lead dissemination efforts in their respective countries in 2022. Each organization will receive full survey results for their specific country as well as infographics and support drafting articles following the survey.

Survey findings should reinforce national advocacy and provide data on experiences of diagnosis around the world to bolster international advocacy. The survey was launched in March 2022.

RDI is part of the survey Expert Committee, which helped develop the questionnaire and will outline a strategy to disseminate survey findings and publications following the survey.

**APEC RARE DISEASE NETWORK**

Established in 1980, the Asia-Pacific Economic Cooperation (APEC) is an intergovernmental forum of 21 ‘economies’ around the Pacific Rim with a seat at the table for industry, academia, and other non-government entities. The primary purpose of APEC is to promote sustainable economic growth, trade and investment, and prosperity in the Asia-Pacific region.
The APEC Rare Disease Network (RDN) was created in 2017 by the APEC Life Sciences Innovation Forum (LSIF) as a tripartite network to address the barriers to diagnosis, treatment, and care of rare diseases in the Asia-Pacific region. More specifically, APEC economies are working together to facilitate greater alignment on national policies and best practices for rare diseases.

The RDN has over 350 members active in 18 member economies. **RDI is part of the leadership of the RDN as a Patient Advisor, represented by Durhane Wong-Rieger.** The RDN helps member economies with the implementation of the APEC Action Plan on Rare Diseases (Action Plan), which was endorsed by APEC Health Ministers in 2018. The objectives of the Action Plan are to:

1. Facilitate greater alignment of domestic policies and regulations;
2. Support urgent implementation of proven best practices;
3. Promote multisectoral collaborations and patient partnerships.

The Action Plan has more than 70 recommendations to achieve 30 targets across 10 pillars, which include: defining rare diseases and orphan products with policies and processes; raising public and political awareness of rare disease issues; and promoting innovative research and development. The Action Plan is translated into Spanish and Traditional Chinese.

The objectives, strategies, and tactics of the RDN in 2021 included:

1. **Accelerated and harmonized** the design, adoption, and implementation of domestic rare disease policies and plans using the APEC Action Plan on Rare Diseases as a guide and by supporting local stakeholders in APEC economies;
2. **Identified champions from government, patient, clinical, and academic communities** and equipped them with tools and opportunities to support domestic rare disease policymaking and planning, and regional knowledge sharing;
3. **Spotlighted domestic barriers** and regional best practices for expanding fit-for-purpose and coordinated regulatory approval pathways, HTA, and funding mechanisms to improve access to orphan medical products;
4. **Strengthened operations**, drive growth, and maintain the mandate and compliance of the APEC RDN.
International Council for Harmonization of Technical Requirements for Pharmaceuticals for Human Use (ICH) brings together regulatory authorities, and the pharmaceutical industry to discuss scientific and technical aspects of pharmaceuticals and develop guidelines. The establishment and recognition of international guidelines, such as Good Clinical Practice (GCP) set common standards on science and ethics, meaning clinical trials conducted in one ICH region can be used by other ICH regions. In 2021, ICH published a Reflection Paper on Patient-Focused Drug Development (PFDD) that identifies key areas where incorporation of the patient’s perspective could improve the quality, relevance, safety, and efficiency of drug development and inform regulatory decision-making.

RDI intends to engage with ICH to facilitate the collection of feedback from RDI regional communities in public consultations. A brainstorming meeting on the strategy of engagement with ICH took place between volunteer Noel Wathion and the RDI Board of Officers to define our specific objectives and our methods of engagement to have an impact on one or two ICH guidelines, such as the one on Clinical Trials and on Patient Engagement.
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RDI MEMBERS
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FOUNDATIONS

RDI ALLIANCE OF COMPANIES
SUPPORT FOR RDI CAPACITY BUILDING

SUPPORT FOR GLOBAL COLLABORATIONS AND PROJECTS
## ANNEXE 1

### Full List of RDI Member Organizations as of December 2021

1. Advocacy Service for Rare and Intractable Diseases
2. Alianza Argentina de Pacientes
3. Aliber–Alianza Iberoamericana de Enfermedades Raras
4. Alliance Maladies Rares
5. Allianz Chronischer SeltenerErkrankungen e.V.
6. Arabic Organisation for Rare Diseases
7. Asia Pacific Alliance of Rare Disease Organisations
8. Asociación de Familiares y Afectados por Lipodistrofias
9. Associacao Brasileira de Enfermedades Raras
10. Associacao Portuguesa CDG e OutrasDoencas Metabolicas
11. Association Anna
12. Association Aux Pas du Coeur
13. Beacon
14. Blackswan Foundation
15. Botswana Organization for Rare Diseases
16. Canadian Organization for Rare Disorders
17. CDH International
18. Child &Youth Care, Zimbabwe
19. China Alliance For Rare Diseases
20. Chinese Organization for Rare Disorders
21. CMTC-OVM
22. Cutis Laxa Internationale
23. Cyprus Alliance For Rare Disorders
24. DEBRA International
25. EAT – Esophageal Atresia Global Support Groups
26. Esperanza
27. EURORDIS – Rare Diseases Europe
28. Fabry International Network

29. FEDER - Federación Española De Enfermedades Raras

30. Federacion Colombiana De Enfermedades Raras

31. Federación Argentina de Enfermedades Poco Frecuentes

32. Federación Mexicana de Enfermedades Raras

33. FEDERG - Federation of European Associations of Patients affected by Renal Diseases

34. Genetic Alliance Australia

35. Genetic Support Network of Victoria

36. Georgian Foundation for Genetic and Rare Diseases

37. Global Alliance of Sickle Cell Disease Organisations

38. Hypopara Norge

39. Illness Challenge Foundation

40. Indian Organization for Rare Diseases

41. Instituto Vidas Raras

42. International Alliance of Dermatology Patient Organizations

43. International Federation for Spina Bifida and Hydrocephalus

44. International Gaucher Alliance

45. International Niemann-Pick Disease Alliance

46. International Patient Organization for Primary Immunodeficiencies

47. International Prader-Willi Syndrome Organisation

48. Japan Patient Association

49. Leukemia Patient Advocates Foundation

50. Malaysian Rare Disorders Society

51. MCT8-AHDS Foundation Inc.

52. Naevus Global

53. National Alliance for Rare Diseases Support - Malta

54. National Organization for Rare Diseases of Serbia

55. Nicolaides-Baraitser Syndrome Worldwide Foundation

56. NORD - Nord National Organization for Rare Disorders

57. Organization For Rare Diseases India

58. Pancyprian Federation of Patients’ Associations and Friends
59. Pespa (Greek Alliance for Rare Diseases)
60. PH Latin Society
61. Philippine Society for Orphan Disorders
62. Pro Rare Austria, Allianz für seltenen Erkrankungen
63. Rare Disease Foundation of Iran
64. Rare Disease Hong Kong
65. Rare Diseases Croatia
66. Rare Diseases Ghana Initiative
67. Rare Diseases South Africa NPC
68. Rare Diseases Sweden (Riksförbundet Sällsynta Diagnoser)
69. Rare Disorders Kenya
70. Rare Disorders NZ
71. Rare Voices Australia
72. Retina International
73. Romanian National Alliance for Rare Diseases
74. Russian Patient Association
75. Thalassaemia International Federation
76. The Ehlers-Danlos Society
77. Timothy Syndrome Alliance
78. Wilhelm Foundation
79. World Alliance of Pituitary Organizations
80. World Duchenne Organization
81. World Federation of Hemophilia
Rare Diseases International
2021 Activity Report