Rare Diseases International (RDI) is the global alliance of Persons Living with a Rare Disease (PLWRD) of all nationalities and across all rare diseases.

RDI’s vision is a world where 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:

- **ADVOCATING** for rare diseases as a global policy priority;
- **REPRESENTING** PLWRD and their families at international institutions;
- **SUPPORTING** the empowerment of RDI Members through a Strategic Engagement Programme.

RDI’s Members include national alliances of rare disease patient organizations, regional organizations, and disease-specific international federations that altogether covered 150 countries, by the end of 2022.

RDI’s strategy is defined by RDI Members and RDI Governance structures (RDI Council and the Council of Officers).
Section One: Strengthen RDI as an Organization

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 nine years of existence: four years of existence with its own legal identity 2019-2022 and five 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/

RDI Council Elections

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

<table>
<thead>
<tr>
<th>Council Member</th>
<th>Organization</th>
<th>Year of Election</th>
<th>End of Mandate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Durhane Wong-Rieger</td>
<td>Canadian Organization for Rare Disorders (CORD)</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 (CHARD)</td>
<td>2021</td>
<td>2024</td>
</tr>
<tr>
<td>Alba Ancochea</td>
<td>Federación Española de Enfermedades Raras (FEDER)</td>
<td>2020</td>
<td>2023</td>
</tr>
<tr>
<td>Kelly du Plessis</td>
<td>Rare Diseases South Africa (RDSA)</td>
<td>2021</td>
<td>2024</td>
</tr>
<tr>
<td>Kin Ping Tsang</td>
<td>Rare Disease Hong Kong (RDHK)</td>
<td>2020</td>
<td>2023</td>
</tr>
<tr>
<td>Ritu Jain</td>
<td>DEBRA International</td>
<td>2021</td>
<td>2024</td>
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</tbody>
</table>
Council elections were organized in April 2022 and all Full Members of RDI had the opportunity to vote online to replace or re-elect the following outgoing Members of the Council:

- **Durhane Wong-Rieger**, Canadian Organization for Rare Diseases (CORD), term 2019 – 2022;
- **Yann Le Cam**, EURORDIS – Rare Diseases Europe, term 2019 – 2022.

A total of six nominations were received and 38 Full Members voted. **Durhane Wong-Rieger**, nominated by CORD, was re-elected with 13 votes. **Yann Le Cam**, nominated by EURORDIS Rare Diseases Europe, was re-elected with 14 votes.

**RDI Council 2022 after the Council elections in April 2022**

**RDI Secretariat**

**Staff**

In 2022, one permanent staff member joined RDI:

- **Dolores Cvitičanin** joined RDI in January 2022 as the **Public Affairs Manager**. Dolores is leading RDI’s global policy priorities, including advocating for a WHO Resolution and organising advocacy and policy events.

Over the last few years, RDI has seen extraordinary **growth in its programmes, secretariat, stakeholder network and engagement, as well as in diversified funding sources**. Over the next years, **RDI will build upon these achievements**, expand its partnerships, and engage even more deeply with its members. As the organization continues to evolve and grow its ambition as a strong global voice, it has become necessary to revisit the organizational structure. In December 2022, Flaminia Macchia ended her work as the Executive Director. The Executive Director role will transition to a Chief Executive Officer position. RDI would like to warmly thank Flaminia for her work and dedication to RDI.

**Internships**

- **Lea Jebali** worked at RDI as an Intern from February to July and from October to December 2022, supporting international advocacy activities with a focus on disability.
- **Prune Regnier Grange** worked at RDI as an intern from May to July 2022, supporting international advocacy activities and funding diversification.
- **Ashley Duane** worked at RDI as an Intern for six months from September 2022, supporting the work on access.

### Volunteers

In 2022, RDI also counted on the generosity of high-profile volunteers and would like to warmly thank them for their time and contribution.

### RDI 8th Annual Meetings

#### RDI General Assembly

The **8th General Assembly took place online on 15 June 2022**.

52 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 2021;
- Financial Report for the year 2021;
- Auditor’s Report for the year 2021;
- Auditor’s Special Report on regulated conventions for the year 2021; and
- Action Plan and Budget for the year 2022.
RDI Membership Meeting

The RDI Membership Meeting is an important moment for consultation and engagement on the organization’s strategy and priorities. Members are given an essential overview of ongoing activities and provide invaluable feedback that shapes RDI’s direction, programmes, and activities in the coming years.

The 8th Membership Meeting, entitled: “What’s Next for the Global Community?” took place online on 16 June 2022 and was attended by 92 participants from Member Organizations calling in from over 30 countries. Following the success of the adoption of the UN Resolution on PLWRD and their families in December 2021, the Meeting was a moment for reflection on the next steps and offered an opportunity for Members to shape key policy and programme areas in the coming years.

The agenda focused on three core areas: the RDI Strategic Engagement Programme, Advocacy Priorities, and Global Access.

The session on the Strategic Engagement Programme was opened by Lisa Sarfaty (NORD, USA) and structured as a consultative workshop on the new Programme. Members received an overview of the Programme and had the opportunity to visit and test a dummy version of the RDI Engage and Connect microsite. They offered valuable feedback on programme priorities as well as the microsite’s layout, accessibility, and relevance.

Dolores Cvitičanin, RDI Public Affairs Manager, led an open discussion on global policy. The session featured a conversation on the impact of the UN Resolution on PLWRD and their families and the next steps towards advocating for Universal Health Coverage and Health Equity for PLWRD within a proposed World Health Organization (WHO) Resolution. The discussion was supported by contributions from Roselyn Odero and Christine Mutena (Rare Disorders Kenya), Agnes Hoctor (International Prader-Willi Syndrome Organisation), Anna Meriluoto (Fabry International Network) and Salome Mekhuzla (World Federation of Hemophilia).
The session on Global Access was facilitated by Mary Wang, RDI Science Policy Manager, and focused on the Operational Description of Rare Diseases, the IRDiRC-RDI Access Task Force, and the Essential Medicines Lists for Rare Diseases. Helen Malherbe (Rare Diseases South Africa), Rachel Yang (Chinese Organization for Rare Disorders), Nuala Ryan (NCBRS Worldwide Foundation) and the WHO Classifications, Terminologies and Standards Unit offered insights on the development of global access initiatives and opportunities for rare disease patient advocates to take part in this area of work.

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

The [RDI Alliance of Companies for Patient-Centred Action]( AoC) provides a platform for companies to support 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 RDI 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 2022, the following **16 companies** joined the RDI AoC: Alexion, Amicus, Biogen, Blueprint Medicines, Boehringer-Ingelheim, CSL Behring, Janssen/Johnson & Johnson, Kyowa Kirin, Novartis, Pfizer, PTC Therapeutics, Roche, Sanofi, Takeda, UCB, and Vertex.

The RDI Secretariat organized **three meetings** with the members of the RDI AoC in 2022, more specifically in May, July, and October:

- **On 24 May 2022,** in Geneva, Switzerland, RDI presented the **Collaborative Global Network for Rare Diseases** to share progress on the programme and obtain perspectives from companies;
- **On 5 July 2022,** online, RDI introduced the **Operational Description of Rare Diseases**, to obtain feedback from companies and discuss the impact from the perspective of the pharmaceutical industry;
- **Finally,** the online meeting on 5 October 2022 focused on **RDI Capacity Building Programme**, to share RDI’s medium-term capacity-building strategy and activities for 2022-2025.
Moreover, RDI shared progress on International Policy, which included information about the #Act4Rare Toolkit and the campaign towards a WHO Resolution.

Section Two: Maintain & Improve Legitimacy

RDI Membership

RDI Members are crucial to building an active national, regional, and global movement to advocate for global policy and drive global initiatives for rare diseases.

In 2022, Members were involved and engaged through the RDI Strategic Engagement Programme and regular communication with the secretariat. Members were also represented in the RDI Policy Committee, RDI Working Groups on Access and the Operational Description of Rare Diseases, as well as the Expert Panel of the Collaborative Global Network for Rare Disease. In addition, RDI facilitates the participation of its members in IRDiRC task forces and working groups.

Starting with 20 Member organizations in 2015, RDI’s membership grew to 86 national and regional organizations as well as disease-specific federations by the end of 2022, gaining five new Members in 2022.

New Members include national alliances from Israel, Burkina Faso, Guinea, and Lesotho, as well as two international federations. Through its membership, RDI is active in 150 countries worldwide and on six continents.

<table>
<thead>
<tr>
<th>New Members in 2022</th>
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| Coalition of Rare Diseases in Israel  
(Joined 13/06/2022) | International Federation of Psoriasis Associations  
(Joined 11/05/2022) |
| Fondation Internationale Tierno et Miriam (FITIMA) - Burkina Faso and Guinea  
(Joined 02/11/2022) | Rare Diseases Lesotho Association  
(Joined 28/01/2022) |
| Fragile X International  
(Joined 24/06/2022) |

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

Outreach and Communication

In 2022, RDI’s Outreach and Communication focused on developing clear and consistent messaging that targets a range of relevant stakeholders, including rare disease civil society
groups, other NGOs for vulnerable communities, international institutions, public bodies, and the private sector. Outreach and Communication reinforce the organization’s position as a well-informed, representative, and global alliance for PLWRD around the globe. RDI has continued to develop a strategy that simultaneously highlights the organization’s core identity as a grassroots alliance while asserting itself as an authoritative voice in global health and social justice.

Editorial guide

RDI has drafted a new editorial guide with a precise lexicon and vocabulary unique to RDI. The guide ensures consistency and coherency across programme areas, activities and various platforms (website, social media, PowerPoint presentations, position papers, editorial articles, and all other communication outputs). It also gives guidelines on RDI’s organizational narrative (how to tell RDI’s story) and provides a specific visual identity which privileges images of the global rare disease community and a vibrant colour pallet.

Key points of the 2022 editorial and style guides:

- Accessible and inclusive language;
- Based on principles and standards developed by the United Nations;
- Promotes the use of “Persons Living with a Rare Disease” (PLWRD) as a term to refer to our community; and
- Has a vibrant and colourful visual identity which highlights the diverse global rare disease community.

Website

RDI website has continued to expand its reach since the new site interface was launched in December 2021. In 2022, the site improved the user experience by simplifying page navigation, providing greater content on programmes and ongoing activities, and improving the quality of translated pages available in French, Portuguese, and Spanish.

The number of users has grown progressively over the last three years, with 13 000 unique users in 2020, 21 000 visitors in 2021, and 26 000 in 2022. Users from the USA and Western Europe represented over 25% of all site traffic; however, the site has gained visitors from Brazil, India, Pakistan, and South Africa.

Social media

In 2022, RDI continued to increase its visibility and following on social media to support international policy priorities and reach the broader rare disease civil society community.
LinkedIn is the fastest-growing social media platform, with 2,725 new followers on the RDI page in 2022. Through LinkedIn, RDI disseminated advocacy positions and calls to action, posted recruitment announcements, and promoted materials on programme areas, including thought pieces produced by the Global Rare Disease Network and resources on the Operational Description of Rare diseases.

Twitter proved an effective tool to promote the activities of RDI’s Members and other rare disease civil society groups, as well as share advocacy priorities and event registration. At the end of 2022, RDI’s Twitter profile had over 4,200 followers and published an average of five individual posts a week.

Since its launch in November 2021 to the end of 2022, the Facebook page has been visited 2,745 times. Facebook extends RDI’s public visibility and helps the organization connect with rare disease organizations and support groups.

**Event Communication and Promotion**

The Outreach and Communication Team developed branding and communication packages for events throughout the year and supported event organization and registration. Communication materials included the design of agendas, concept notes, registration pages and social media content.

Materials were developed for the following 2022 events: RDI Membership Meeting on 16 June 2022; [RDI Side-Event on the Margins on the 75th WHO World Health Assembly (WHA)](RDI Side-Event on the Margins on the 75th WHO World Health Assembly (WHA)) on 24 May 2022; [Formal Side-Event to the UN High-Level Political Forum on Sustainable Development (HLPF)](Formal Side-Event to the UN High-Level Political Forum on Sustainable Development (HLPF)) on 6 July 2022; and [UHC Day 2022 – Universal Health Coverage for Rare Diseases](UHC Day 2022 – Universal Health Coverage for Rare Diseases) on 12 December 2022. The team also developed the microsite and provided creative direction for “[Rare Diseases: A Global Priority for Equity](Rare Diseases: A Global Priority for Equity)” which took place on Rare Disease Day 2022 at Expo Dubai.

**Media Highlights**

- [Ensuring children with a rare disease are no longer left behind](Ensuring children with a rare disease are no longer left behind, Health Awareness)
- [Out of the Darkness, Rare Resolution Magazine](Out of the Darkness, Rare Resolution Magazine)
- [Transforming care for rare diseases around the world, Health Awareness](Transforming care for rare diseases around the world, Health Awareness)
Section Three: Strategically Improve Capacities

RDI Strategic Engagement Programme

In 2022, the Capacity Building Programme became the Strategic Engagement Programme to reflect a more holistic approach to the engagement of members and patient advocates and builds RDI's capacity to act as a global alliance with an active grassroots base. The Programme captures the organization’s medium-term capacity-building strategy and activities for 2022-2025.

Programme development is informed by a two-year consultation process which includes the assessment of organizational capacities and resources, a survey of the Membership, one-on-one discussions with Members on capacity-building needs, and feedback and consultation with the RDI Council. The outcome is a comprehensive Programme which aims to strengthen the capacities of rare disease civil society groups and patient advocates while supporting RDI’s capacity to act as a representative voice for the international rare disease movement.

Programme Impact

In addition to offering toolkits and resources, opportunities for international networking, regional dialogues, and knowledge-sharing, the Programme also supports more effective collaboration with patient groups in global initiatives and global policy actions.

RDI Strategic Engagement focuses on three levels of impact: RDI Member organizations (the main target of programme activities), individual rare advocate leaders, and the broader rare disease civil society community.

Programme Objectives

- Support the empowerment of rare disease patient groups to drive action and advocacy at local, national, and regional levels.
- Raise engagement and enable the broader community to shape international collaborations and policy development.
- To support individual rare disease community leaders.

Programme Priorities

The Programme is structured around three priority areas: Policy, Global Collaborations, and Access and Health Equity. The three priorities were identified through the comprehensive consultation process and form distinctive learning paths within the programme.
The three paths

- **Policy and advocating for Persons Living with a Rare Disease and their families** - focuses on international policy action advocacy with particular attention on RDI policy campaigns, toolkits, and calls to action. The path also supports patient advocates to shape national plans and frameworks through resources such as a list of national plans and definitions and small group meetings on advocating for national frameworks.

- **Global Collaborations** - focuses on existing and emerging international partnerships driven by RDI or where RDI and its members are active and have a seat at the table to help drive progress. The Programme also disseminates information and learnings developed within various international rare disease collaborations.

- **Global Access and Equity** - focuses on providing resources on regional rare diseases access models, orphan drug legislation, as well as international programmes supporting health equity.

Programme Element and Activities

**RDI Engage & Connect**

The *Strategic Engagement Programme’s activities and resources are available on a microsite within the RDI website entitled - RDI Engage & Connect*. The centralised hub enables visitors to follow three learning paths depending on their interests. It also allows users to download the most relevant open-source resources and materials developed by different stakeholder groups, including other patient groups. In addition, RDI-generated toolkits, calls for fellowships, and a calendar of events and conferences can be accessed on Engage & Connect.

Our Members tested a trial version of the Engage & Connect platform at the RDI Membership meeting on 16 June 2022. Member feedback on the platform’s usability, layout and content was collected through live polls and discussions. It led to changes in the microsite design, accessibility and content before officially launching in October 2022.

Issues that still need to be addressed include providing materials in more languages, improved filtering and sorting of resources, and diversification of the types of resources on the site.

**RDI Regional Conferences**

*Regional Conferences are organised by Members in partnership with RDI* to facilitate dialogue on issues specific to regions and help nurture regional collaborations and initiatives. As a partner to regional conferences, RDI plays a role in programme committees, helps identify speakers, and provides a limited budget to cover event planning.
Regional Conferences in 2022:

- **Asia Pacific Alliance of Rare Disease Organisations (APARDO) Conference, Thailand, 25 – 27 November 2022**

APARDO’s annual meeting welcomed over 100 precipitants from 98 countries. It covered a range of relevant topics, including newborn screening and genetic testing, advances in policies and programmes in the Asia Pacific region, and rare disease registries driven by patients and patient groups.

APARDO’s Conferences gather multiple stakeholders to discuss and advance issues relevant to the region and provide an important opportunity for patient advocates to share learnings and build support networks. The 2022 agenda was composed of facilitated workshops, panel discussions, roundtables, and presentations from Persons Living with a Rare Disease and patient advocates from across the region.

Universal Health Coverage (UHC) and ensuring better health for all were key to the discussion, which included a workshop facilitated by RDI’s Public Affair Manager, Dolores Cvitičanin. The workshop collected feedback and contributions from the region, which will be integrated into the final document presenting civil society’s key asks for a proposed WHO World Health Assembly Resolution on UHC for rare diseases.

Through the Strategic Engagement Programme, RDI was a partner to the event, contributing to discussions on UHC and awarding APARDO a small grant toward event logistics.

- **Africa Alliance Meeting, Kigali, Rwanda, 28 – 29 November 2022**

The African Alliance Meeting was an intensive two-day gathering of African civil society leaders to establish an African Rare Disease Alliance and bring together regional rare disease groups to encourage collaboration and strengthen the continent’s movement.

The gathering was hosted by Centre Alliance for Rare Diseases - a non-profit organization working to improve the lives of children with a rare disease in Rwanda. It was co-organized by the following African groups: Ali Kimara RD Foundation – Tanzania, Association Aux Pas Du Coeur - Cote d’Ivoire, Botswana Organisation for Rare Diseases, Cardiac Community Nigeria, Child and Youth Care Zimbabwe, FITIMA - Guinea Conakry, Mali Rare Disease Association, Multiple Sclerosis Namibia, Rare Disorders Kenya, Rare Diseases Ghana Initiative, Rare Diseases Lesotho Association, Rare Diseases South Africa, Rare Disease Uganda, Rare Diseases Nigeria, Taxawuma - Senegal Rare Diseases, and Rare Diseases & Disabilities Africa Foundation - Zimbabwe.

The meeting followed a previous conference in South Africa (2017) and a summit held in Ghana (2021). Participants outlined the vision, mission, governance structure, and sustainability model for the proposed regional federation for rare diseases.
The RDI Strategic Engagement Programme supported the event through participation in weekly planning meetings, helping to draft meeting documents, presenting a global perspective at the event, and awarding a grant towards event logistics.

- **Iberoamerican Alliance for Rare Diseases (ALIBER) 2022 Annual Meeting, Costa Rica, which was postponed to 9 – 10 February 2023**

The ALIBER Annual Meeting is a platform for patient leaders from the Iberoamerican region to collaborate and host cross-stakeholder conversations.

In 2022, the RDI Strategic Engagement Programme awarded a grant to ALIBER to assist with event logistics. RDI also facilitated a pre-conference session led by Jesus Navarro (Organización Mexicana de Enfermedades Raras - OMER) and Roberta Anido de Pena (Federación Argentina de Enfermedades Poco Frecuentes - FADEPOF). The online pre-conference meeting collected feedback and contributions from the region which will be integrated into the final document detailing civil society’s key asks for a proposed WHO World Health Assembly Resolution on UHC for rare diseases.

Due to challenges securing the meeting space in 2022, the event was postponed and took place from 9 to 10 February 2023.

**RDI Fellowships**

In 2022, the fellowship programme was incorporated into the broader RDI Strategic Engagement Programme. Fellowships offer travel and accommodation support to enable patient advocates to attend workshops, meetings, and conferences worldwide. Through RDI fellowships, patient advocates can build a network of support which includes patient leaders and other stakeholders, take part in discussions shaping the rare disease context, and build organizational capacities.

**Fellowships awarded in 2022:**

- **European Conference on Rare Diseases, online, 27 June – 1 July 2022**

With support from EURORDIS, RDI awarded 20 free places for patient advocates to attend the 11th edition of the ECRD entitled: "Mission Possible - putting rare disease policy in action". ECRD fee waivers were awarded to patient leaders from - Australia, Bulgaria, China, Colombia, Colombia, Egypt, Georgi, Kenya, India, Malaysia, Mexico, Pakistan, Rwanda, Tanzania, and the USA.

- **World Orphan Drug Congress, USA, Boston, 11 – 13 July 2022**

In 2022, RDI granted travel and accommodation support to the following patient leaders: Fernando Gil Cardozo (Federación Colombiana de Enfermedades Raras - FECOER), Amira...
Awada (Vidas Raras - Brazil), Miriam Ramírez Lopez (Association of Urea and Metabolic Cycle Disorders Peru) and Nthabeleng Ramoeli (Rare Disease Lesotho Association).

Following the event, fellows submitted a survey on the value of attending the conference and individual learning.

“Thanks to you, we contacted industry professionals. We hope they will want to bring their technologies to Peru” Miriam Ramírez Lopez

“The most important thing was networking. We met with different role players in the rare disease space.” Nthabeleng Ramoeli

- **World Orphan Drug Congress - Europe, Barcelona, Spain, 11 – 17 November 2022**

In 2022, RDI granted events passes and travel and accommodation support to the following patient leaders: Fernando Gil Cardozo (Federación Colombiana de Enfermedades Raras - FECOER), Trudy Nyakambangwe (Child and Youth Care Zimbabwe), Zhana Chokheli (SCN2A - Georgia), Rose Okoma (Association Aux Pas Du Coeur - Cote d’Ivoire), Nadiah Hanim Abdul Latif (Malaysian Rare Disorders Society). FECOER received support to attend both the WODC USA and WODC Europe after an awardee withdrew from the programme. However, RDI prefers to support different fellows for each event.

Following the event, fellows submitted a survey on the value of attending the conference and individual learning.

“Attending the WODC was a great experience for me. I think there are lots of other patient representatives who really want to attend the congress, but they can’t afford it.” Zhana Chokheli

“This event allows organizations to see the evolution of therapeutic research, the costs of drug development, and also to understand that there are treatments for orphan diseases available.” Rose Okoma

- **Other RDI Fellowships**

Fellowships for **Africa Alliance Meeting in Kigali** were also awarded to the following RDI Member Organizations: Association Aux Pas Du Coeur - Cote d’Ivoire; Botswana Organisation for Rare Diseases; Child and Youth Care Zimbabwe; FITIMA - Guinea Conakry; Rare Disorders Kenya; Rare Diseases Ghana Initiative; and Rare Diseases Lesotho Association.

An additional grant for fellowships was awarded to APARDO to enable patient advocates from the region to attend **APARDO Conference in Thailand**.
RDI Toolkit and Resources

In 2022, RDI toolkits on the UN Resolution, Universal Health Coverage, and the Operational Definition of Rare Diseases were uploaded to the Engage & Connect platform.

- **#Act4Rare - Toolkit on the implementation of the UN Resolution**

The Strategic Engagement Programme supported the development and design of the #Act4Rare Toolkit, an initiative led by the Public Affairs team. The Programme designed graphic elements, ensured that materials were accessible and tailored for patient organizations, and oversaw the translation of the toolkit into French, Portuguese, and Spanish.

Resource Bank

On the Engage & Connect platform, RDI collated and disseminated existing materials and tools developed by other patient advocacy groups and international institutions. RDI offers essential lists of free-to-use materials on each of the three programme learning paths (Policy, Global Collaborations and Access and Equity).

Member-led Discussions

Member-led small meetings allow patient advocates to share knowledge and experiences. **RDI Members host meetings which gather between 10 and 15 participants to explore an issue of common interest to patient advocates.**

**In 2022, RDI facilitated two Member-led Discussions:**

- **Advocating for a National Plan** - hosted by Rare Voices Australia.
- **Start a Patient Registry** - hosted by the NCBRS Worldwide Foundation.

Member Feedback and Next Steps

At the Membership Meetings, participants requested that RDI form a working group in 2023 to guide the development of the Strategic Engagement Programme based on learnings from existing rare disease capacity-building programmes and member priorities.

It was also decided that a task force should be formed to assist with reviewing, collating, and translating programme resources.
Section Four: Global Policy Priorities

RDI Policy Committee

The Policy Committee (previously called the “Advocacy Committee”) is responsible for developing and recommending advocacy priorities and policies, as well as preparing RDI position papers, statements, and declarations. The Policy Committee is managed by RDI Public Affairs Manager, Dolores Cviticanin.

RDI POLICY COMMITTEE

In 2022, the Committee convened virtually three times (April, September, and December), with further consultations via email. The Committee contributed extensively to RDI’s Side-Event to the World Health Assembly (WHA) in May 2022, the Key Pillars of Universal Health Coverage (UHC) for Rare Diseases, and RDI’s Universal Health Coverage (UHC) Day virtual event.

Finally, in 2022, the Policy Committee changed its composition. Two members left in November 2022 - Sanja Peric (Rare Diseases Croatia) and Simone Boselli (Eurordis) - as their terms were complete. In a bid to achieve greater regional equity, the two new positions were filled with representatives from Africa and Oceania. Representing Rare Disorders Kenya and Rare Disorders New Zealand, Christine Mutena and Michelle Arrowsmith, respectively, joined the Committee in November 2022. Additionally, in June 2022, Elvira Martinez (FEDER) took over that organization’s position in the Policy Committee from Alba Ancochea. The Committee currently has 18 members, and a Chair (Yann Le Cam - Eurordis).

RDI warmly thanks the Members of the Policy Committee for their contribution and time.
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 and access to care for PLWRD and their families.

As a coordinating partner to Rare Disease Day, RDI promotes the participation of the international rare disease community outside the EU and Northern America and leverages the campaign to advance global policy messages. RDI is an active member of the RDD Steering Committee and takes part in regular Outreach Meetings, giving input on campaign materials and calls to action.

RDI supports the participation of National Alliances (groups working across all rare diseases at the national level) as Official Partners of the RDD campaign. RDI Member national alliances for Cote d’Ivoire and Israel joined the list of RDD official partners in 2022.

On 28 February 2022, entitled - “Rare Diseases: A Global Priority for Equity”, the Fourth High-Level Meeting of the NGO Committee for Rare Diseases took place at Expo Dubai (the World Expo) and online. The hybrid event, organised with RDI, EURORDIS, and Ågrenska, marked Rare Disease Day 2022 and celebrated the UN Resolution on PLWRD and their families. 34 civil society partners also contributed to the gathering, emphasizing the tremendous grassroots movement behind the adoption of the UN Resolution. 60 in-person participants joined us in Dubai, while over 730 attendees tuned in online from 104 countries across the globe.

Presentations from the Core Group of Members States promoting the UN Resolution (Brazil, Spain, and Qatar) as well as Queen Silvia of Sweden highlighted the growing recognition and visibility of rare diseases as a national priority. In addition, strong messages of commitment to the community were expressed through three major UN agencies: the World Health Organization (WHO), the United Nations Office of the High Commissioner for Human Rights
(UNHCR), and the United Nations Educational and the Scientific and Cultural Organization (UNESCO).

Event goal

- Celebrate the adoption of the UN Resolution and outline a roadmap for its implementation at global and national levels;
- Explore international collaboration and synergies needed to make rare diseases a policy priority; and
- Discuss the future of rare diseases beyond health, with a focus on Africa and low-and-middle-income countries.

Key resources

- Meeting recordings and highlights
- Full event report

Rare Diseases and Universal Health Coverage

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.

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.

To celebrate the UHC Day 2022, RDI hosted a webinar with the support of the NGO Committee for Rare Diseases and the UN Office of the High Commissioner for Human Rights on 12 December. During the webinar, expert speakers proposed actions needed to make UHC a reality for all. Moreover, rare disease civil society groups in Africa, Latin America, Asia, and Europe shared successes and good practices in their regions since the
adoption of the UN Political Declaration on UHC (2019) and the UN Resolution on PLWRD and their families (2021).

**Rare Diseases and Disability**

In January 2022, RDI launched a body of work to understand the commonalities and differences between rare diseases and disabilities, what challenges and impacts they may present, and how advocacy can be conducted so that the two communities could work together towards common goals. This work was led by RDI’s Public Affairs Intern, Lea Jebali.

As part of the body of work, a 41-page document, entitled “Commonalities and differences between rare diseases and disabilities: Challenges and impacts”, was produced, which employed a sequential, exploratory, and mixed-method design to identify differences and similarities. Data was generated through a total of 30 expert interviews, including with 23 rare disease experts and seven disability experts, and followed by a quantitative RDI Member survey. The document concludes by stating that:

> It is important to recognize the commonalities and differences to understand how the different dimensions function and influence each other. As a practical contribution, the rare disease community and the disability community can benefit from an intersectional understanding to promote change that includes Persons with Disabilities and Persons Living with a Rare Disease.

RDI also developed an “Advocacy Strategy for rare diseases and disabilities”, to form a strategic partnership between the two communities. The strategy’s vision is to achieve intersectional justice for rare diseases and disabilities by working together on the commonalities and independently on the specificities.

On 19 July 2022, RDI hosted the webinar event “Rare Diseases and Disabilities”, during which the two aforementioned documents were presented. Participants from civil society discussed the findings and considered how to best move forward with the advocacy strategy.

In the last quarter of 2022, RDI executed elements of the advocacy strategy and set up a protocol for its further implementation in 2023.

**Act for Rare**

In 2022, RDI created the #ACT4RARE Toolkit. Containing a stakeholder analysis grid, a template letter to a policy maker, and a comprehensive social media kit, amongst other elements, this Toolkit is designed to empower patient advocates around the world to demand national action on the UN Resolution on “Addressing the challenges of persons living with a rare disease and their families.”
The 30-page Toolkit, available in English, Spanish, and French, was launched via a webinar on 12 October 2022. During the webinar, the RDI Secretariat introduced the toolkit, and rare disease advocates spoke about how they planned on utilising it.

**NGO Committee for Rare Diseases**

The NGO Committee for Rare Diseases aims to promote multi-stakeholder collaboration and actions for rare diseases within the United Nations system. It was established in 2015 under the umbrella of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO) and acts as a forum of interested parties such as NGOs from the field of rare diseases and beyond, United Nations bodies and agencies, as well as individual experts.

To mark Rare Disease Day 2022, the NGO Committee for Rare Diseases co-hosted a high-level policy event at EXPO Dubai with RDI, the Ågrenska Foundation, and EURORDIS-Rare Diseases Europe. The event, entitled “Rare Diseases – A Global Priority for Equity”, marked the first major gathering of the global rare disease community since the adoption of the historic UN Resolution. The event convened stakeholders to discuss how this UN Resolution could drive greater equity for PLWRD and ensure that PLWRD enjoy an improved quality of life.

The NGO Committee for Rare Diseases also acted as a Co-Organizer of RDI’s Formal Side-Event to the High-Level Political Forum (HLPF) in July 2022. “Building back better from Covid-19 and advancing the 2030 Agenda: Addressing the challenges of Persons Living with a Rare Disease as a gender equality, human rights, and equity priority” featured multi-stakeholder speakers touching on the intersection between Sustainable Development Goal (SDG) 5 - Gender equality - and rare diseases.

**Section Five: Participate in / lead Global Initiatives**

**Memorandum of Understanding between WHO and RDI: Third year of execution**

**Operational Description for Rare Diseases**

In order to improve the recognition of rare diseases and their visibility in health systems, the WHO and RDI agreed to collaborate towards an Operational Description of Rare Diseases to be considered when shaping policies that impact on the lives of PLWRD and their families. In the second year of the Memorandum of Understanding (MoU), RDI, in collaboration with a global panel of rare disease experts, developed an internationally endorsed [Operational...](#)
**Description of Rare Diseases**, which was submitted as a deliverable to the WHO (March 2022). The reference document enables a uniform, objective interpretation of what diseases are considered rare, how many persons are affected and why the defined rare disease population demands their attention. The activities in the third year of the MoU focused on performing an impact assessment on the adoption of the reference, as well as communication and dissemination of the document.

The impact assessment with a wider group of stakeholders suggests the adoption of the document would provide opportunities in research and development, prioritization and organization of essential health and social care services for PLWRD. The impact assessment was accomplished through three online workshops with a range of global stakeholders. More than **55 participants from 47 organisations** with expertise on research and development activities and health and social care services for PLWRD contributed to these workshop discussions. Participants came from RDI membership (9); funding agencies, research organizations, hospitals (12); international/regional consortia (6), regulatory agencies (3), and private sector (including RDI AoC).

The collaboration with WHO and the alignment of the core definition concepts with the International Classification of Disease (ICD) was a fundamental step in supporting better data registration and visibility of PLWRD in health systems. On **27 September 2022**, WHO Classifications and Terminologies Unit in collaboration with the Healthier Populations Division, co-hosted a webinar with RDI and Orphanet on the use of ICD-11 for rare diseases. The webinar was attended by 1151 participants from 113 countries.

The reference document was presented during the RDI policy event during the 75th WHA and as a poster at the European Conference on Rare Diseases 2022, which was awarded as the 3rd best scored poster at the conference. RDI supported a roundtable discussion on the subject of harmonising an international definition at the World Orphan Drug Conference USA, Boston, July 2022.

At the end of 2022, RDI began developing an animation video titled “**What is a Rare Disease?**” to explain the Operational Description of Rare Diseases for the broader audience. This video aims to complements the previous video produced by RDI “**Life with Rare Disease**”. Supporting postcard and infographic were made available on the dedicated RDI webpage.
Global Rare Diseases Network

RDI has continued the development of the Work Programme, supported by the Technical Research, in order 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. RDI collaborates with the WHO to prepare for the launch of a pilot phase of the network.

The work to be performed during year 3 of the MoU focuses on the mobilisation of the rare disease community, successfully complete the conceptualisation and technical research, moving into the next stage of action. The framework for the network is based on the agreed strategic bottom-up and top-down approach that promotes the identification of specialised multi-disciplinary centres of expertise, connects them regionally and internationally into the Global Network, while encouraging local capacities to generate additional expertise.

Technical Research Progress

The Technical Research in 2022-23 was conducted by RDI and its member Patient Organisation Members, in partnership with a Panel of Experts from around the globe.

1. Concept Model for Rare Disease Hubs including suggested requirements for governance, and the mandate/mission. A Sustainability Working Group was formed in January 2022, with 17 experts from 17 countries and met regularly throughout 2022. The RDI Team conducted a desktop review of the existing national, regional, and global collaborations and networks to identify common structures and functions. The vision of the global network, set out in the Concept Model, proposed the connection of existing collaborations, networks and communities under a ‘hub and hubs’ model. The Rare Disease Hub model was developed based on guiding principles that enable local communities to define and tailor their participation based on their local policies and infrastructure and anchored into the local system.

2. Horizon scanning on earlier adopters and innovators for a list of potential Network Participants. The RDI Team, through engagement of the rare disease community in a series of regional online workshops, tested the ‘readiness’ of centres, communities and
countries to participate in the up-and-coming Pilot Network. Readiness was appraised based on three core areas: i. willingness to participate and collaborate; ii. existing infrastructure or networking to build from; and iii. potential to secure endorsement from the healthcare provider, institution and/or government. The baseline appraisal was presented to each region to secure feedback and for further elaboration. In these workshops, the communities determined the key actions needed to move from engagement into action, identified potential participants for the WHO Pilot Network, and mapped the strengths, experience and potential added-value of each one. In 2022, the team delivered two Phases of online workshops and webinars to support the communities to prepare for the Pilot network. Workshops in Phase I, Q1-2, supported the regional collaborations and discussions of hub functions. A total of 80 experts (33 patient representatives and 47 clinicians) from 37 countries participated in these workshops. Six regional workshops were organised with the Panel of Experts and held online in Q3 2022 to support the development of applications from WHO Regions. A total of 84 experts (39 patient representatives and 45 clinicians) from 35 countries took part in these workshops. The main findings from the workshops have been developed into deliverables.

3. Updated Digital Specification for the Pilot Network, scoping the requirements and a horizon scanning of possible solutions.

a. A literature review of the evidence for virtual care: The RDI Team conducted a targeted literature review on ‘virtual care’ to draw out the important features that would best serve and benefit a global network for rare diseases. The literature was large in volume, broad in scope and currently poorly organised in terms of frameworks, terminology, and concepts. The findings highlighted that virtual care is best understood as ‘remote care’ and ‘networked expertise’.

b. Mapping exercise of the function requirements and existing tools: In preparation for the Pilot Network of the GRDN, a Digital, Data and Technical (DDT) Working Group was established in Q4 2021, following expressions of interest from members of the Panel of Experts. A total of 26 members from 21 countries joined the DDT Working Group. The overarching objective of the DDT Working Group was to identify effective, user-friendly platforms and tools that support national, regional, and global collaboration of the members of the Pilot Network. The DDT Working Group was co-chaired by two members of the Panel of Experts who are international leaders in both rare diseases and e-health. The DDT Working Group met online regularly throughout 2022. Through a series of online workshops, the DDT working group mapped the clinical needs for networked care – networked expertise, remote care, learning and information sharing - and subsequently quantified the digital, data and technical requirements associated with each of these functions. A survey of the Panel of Experts was completed to map how ‘Virtual Advice And Consultation’ (VAAC) within countries and across regions was currently managed. The survey received a total of 29 responses from 22 countries.
Semi-structured interviews were held with 12 experts from different settings – low-middle- and-high income regions - to complete a deep-dive into existing VAAC systems. A series of ‘use cases’ of existing VAAC platforms were developed. The report and recommendations were completed based on the activities of the group.

**Communication Activities**

The RDI Team has developed a set of communication tools and materials, available in different languages, to be used by rare disease community leads to engage with their Expert Centres and Health Authorities or National Policy Leads to raise awareness and interest around connecting to and participating in the Pilot Network.

**RDI Policy Event as an informal side-event to the WHA (24.05.2022)** - The event presented recommendations for strengthening healthcare systems for PLWRD, with an in-depth look at the creation of a GRDN. The event shared the proposal and discussion of five concrete actions for strengthening healthcare systems. Finally, a panel presented added-value of networking care globally to establish a bottom-up approach to strengthening healthcare systems for PLWRD. It was a historic moment: the launch of the ground-breaking collaboration between RDI and the WHO toward a GRDN.

**Global Network Workshop with Panel of Expert Leaders (25.05.2022)** - The workshop took place in Geneva, Switzerland, following the RDI Policy Event in the context of the 75th WHA. The workshop was a closed meeting of representatives from the Panel of Experts of the GRDN, from 13 different countries. Of the fourteen experts, nine were patient representatives and six were clinicians, representing the six world regions from the WHO. The workshop was structured into two sections – strategic approach and operational actions. Attendees believed that capturing the support of the WHO with the needs of the rare disease community, in one voice, would facilitate action locally in each country.

**External Communication Campaign** - Launched in September 2022, the RDI Team initiated a plan to publish monthly articles from Panel of Experts members that build understanding around networking, pooling of expertise and collaboration, as effective and sustainable ways to meet the needs of PLWRD. The articles were developed through interviews with experts from the Panel of Experts to support the rationale of networked care, promote the key benefits, delineate why this is the way forward for rare diseases, and share examples of current collaborations. Five articles were published. Each article is displayed on the RDI website and included in the RDI Newsletter. RDI social media channels have been leveraged to feature the articles on LinkedIn and Twitter.
WHO Model List of Essential Medicines

The third area of collaboration with WHO promote access to health services via the facilitation of patient access to essential medicines and essential in vitro diagnostics. More specifically, the objective is to explore how a reference list of medicines and diagnostics for rare diseases could provide Member States with a model from which develop a national priority list and inform policies. To explore the concept of such a model list for rare diseases, collaboration was created 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 during the second year of the MoU. In the third year of the MoU, RDI and WHO Model List Units (EML, EDL) were engaged in conversation, to validate the gap analysis of the different mappings of essential medicines for rare diseases, on the outlook regarding capacity building, to take a first evaluation of EDL for rare diseases.

**Validation of mapping efforts** - Many medicines in the WHO EML and EMLc have indications for common conditions, even if they also have regulatory approval for treatment of rare diseases. A working proposal was shared with the WHO and discussed, leading on to exchanges of respective mapping lists of rare disease medicines in EML and EMLc. This list of medicines compiled by the EML Secretariat was shared with RDI for comparison with the IRDiRC and 2021 Deliverable Lists. The analysis on the respective mapping efforts and the documentation of the differences suggested that further dialogues would support a common interpretation on how to capture all the medicines relevant for rare diseases in the EML and EMLc. The EDL was evaluated within the same goal, to facilitate access to essential in vitro diagnostics. Differing to EML in which each medicine is listed with its treatment indication or disease, IVDs are not always associated to a specific disease. Most IVDs provide a biomarker reading or provide a diagnostic result which indicate further specialized tests. In considering that the aim of EDL and EML is to complement each other, it would be an opportunity to explore the complementarity of the Model Lists for rare diseases.

**Engagement with the rare disease community.** In 2022, RDI launched several discussions and engagements with regards to rare disease essential medicines in several international conferences to a wide geographies and range of stakeholders, including RDI membership. These engagements ranged from online workshops, oral presentations at conferences, poster and webinars: RDI webinar for members on 30 March 2022: European Conference on Rare Diseases; the International Rare Disease Showcase; World Orphan Drug Congress; Symposium on Birth Defects and Rare Disease; and Africa Summit on Rare Diseases.

**Responding to WHO public consultations**

**Accelerating Access to Genomic Technologies:** RDI provided a feedback to the WHO Science Council for their recommendation report for the WHO Director-General on accelerating
access to genomic technologies in low- and middle- income countries in April 2022. The feedback was formulated via a small focus group with RDI members and experts. The key messages in the feedback were made available on the RDI website. The WHO Science Council recommendation report was published in July 2022.

**Global regulatory framework for cell and gene therapy products:** RDI provided a written comment on the white paper **WHO approach towards the development of a global regulatory framework for cell and gene therapy products**. The white paper was analysed with an expert volunteer in regulatory science and forwarded to the WHO Expert Committee on Biological Standardization (ECBS).

**Collaborations with IRDiRC**

The International Rare Disease Research Consortium (IRDiRC), 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 is governed through a Consortium Assembly, an Operating Committee, three Constituent Committees and four Scientific Committees, aided by ad hoc Task Forces. In 2022, RDI took part and contributed to a series of meetings within the following IRDiRC structures: Consortium Assembly, Patient Advocates Constituent Committee (PACC), and numerous Task Forces. RDI represented by Durhane Wong-Rieger, as Chair of the PACC, and contributes to the Operating Committee.

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. RDI members and patient representatives are contributed to all of the IRDiRC Task Forces which were activated in 2022. Of note, many RDI members were nominated to the Task Force “**Enabling and Enhancing Telehealth for Rare Diseases Across the Globe**”. The RDI Secretariat has contributed to the **Primary Care** and **PLUTO Project** Task Forces. The Primary Care Task Force seeks to identify the priority research areas in primary care that need to be addressed to deliver against the IRDiRC goals, to identify current state of play, and identify challenges and opportunities in rare diseases research in primary care. The PLUTO project aims at using an integrated database search approach to: identify and classify the groups of rare diseases that are currently under-represented by academic research and industrial development alike and to determine what characteristics they have in common.

A new task force proposal led by Durhane Wong-Rieger and the PACC on **“A framework to assess impacts associated with diagnosis, treatment, support, and community integration that can capture changes along the rare disease patient and family journey”** (Impact
Framework) was prepared and approved by the IRDiRC Consortium Assembly for activation. The new Impact Framework Task Force will recruit experts and be operational in 2023.

RDI took part in the IRDiRC and EJPRD organised International Conference on Clinical Research Networks for Rare Diseases, 1 – 2 December 2022, Paris, and contributed with a presentation on “RDI-WHO Collaboration toward Global Rare Disease Networks”.

Global Access to Medicines

RDI-IRDiRC Global Access Working Group

The IRDiRC Rare Disease Treatment Access Working Group compiled the first Essential List of Medicinal Products for Rare Diseases in 2021 with the goal to improve the standards of care. The newly restructured RDI-IRDiRC Global Access Working Group (WG) since November 2021 underwent a review to enable a shift to focus on the second phase of the objectives, to initiate research into barriers to accessing essential rare disease medicines. There are 20 members in the Global Access WG, including eight RDI members.

The Working Group adopted a case study approach for the studies. A bottom-up approach was taken to gather real experiences from stakeholders (patients, patient advocacy groups, physicians). The first case was on Cystinosis, a lysosomal storage disease. The structure of the case study consisted a brief literature review, mapping of available medicines and stakeholders for the condition. Interviews were organised and conducted by Dr Mary Wang with findings provided regularly to the WG for discussions. Interviews were carried out in 11 countries for statements regarding access: Australia, Canada, Colombia, Ghana, Malaysia, Mexico, New Zealand, South Africa, Spain, Thailand, Vietnam. The access to cysteamine for cystinosis case is a good example to illustrate that challenges along the steps of patient journey should be addressed and a tight relationship between availability of diagnostic capabilities with access to medicines. The second case on Cystic Fibrosis began in October 2022 with the starting desk research carried out by the Global Access Intern.

Global Alliance for Rare Disorders Access (GARDaccess) project

Partnership for Quality Medical Donations (PQMD), a unique alliance of 44 non-profit and corporate member organizations committed to bringing measurable health impact to underserved and vulnerable people through active engagement with global partners and local communities, launched an initiative called the Global Alliance for Rare Disorders Access (GARDaccess). The mission of GARDAccess is to accelerate patient access to quality medicines, treatments, and services for rare diseases by harnessing the collective expertise of partners with shared values. There were two workstreams: to develop quality guidelines for rare disease therapeutics and medical support, and to develop a framework to ultimately implement a sustainable model for rare diseases. RDI took part in the Workstreams in the of
the initiative and Durhane Wong-Rieger participated as a panelist in the PQMD Pillar Talk Webinar on GARDaccess. The outputs of the GARDaccess are available on the dedicated website. The Phase I of the initiative concluded at the end of 2022 and RDI is in discussion with PQMD on furthering a collaboration for Phase II.

## Diagnosis

### Global Commission and Rare Barometer Survey

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease is a multidisciplinary group of experts convened by EURORDIS, Takeda and Microsoft. With the Global Commission, RDI advances equitable access to diagnosis, focusing on regions with limited pathways to rare disease diagnosis.

In June 2022, the EURORDIS Barometer Programme, working closely with RDI and supported by the Global Commission, launched “The Journey to Diagnosis for People Living with Rare Diseases” - a global survey collecting data on experiences of diagnosis for PLWRD and their families around the world. The survey was translated into 27 languages, and 13,300 respondents took part from 107 countries worldwide.

RDI led outreach and engagement to six pilot countries: Argentina, Australia, Brazil, Malaysia, South Africa, and the United Arab Emirates and ensured the translation of the survey into Arabic and Malay. RDI’s partners in the six pilot countries received detailed survey results, which will help to strengthen national advocacy action on the diagnosis of rare conditions. RDI will use the findings of the global survey to support policy statements on accelerating diagnosis and elevating the barriers to timely diagnosis.

### IRDiRC Newborn Screening

IRDiRC has launched a discussion on the importance of Newborn Screening (NBS) with a focus on covering the topic by including examples of implementation, usage of results, ethics, and future state. As different rules and regulations apply in different countries, IRDiRC sought to bring the contribution in articulating the status of the science and its applications in NBS. Ideas for two thematic journal collections were launched: “Real World Applications and Technologies”, and “Policy, Ethics and Patient Perspectives”. RDI has agreed to contribute to the second special edition. Several online meetings took place in 2022 to discuss and agree on the scope of the collection, gap analysis, reviewing of topics of interest.

## Alignment on Global Agenda

IFPMA
In 2017, International Federation of Pharmaceutical Manufacturers & Associations (IFPMA) published a position paper on a set of policy principles and organised a roundtable to facilitate discussions regarding rare disease challenges. At World Orphan Drug Congress, USA, RDI shared a panel discussion with IFPMA to hear IFPMA’s commitment to advocate for more patient-centric policies and robust health systems to bring innovation to rare disease patients at the multilateral level. The new interaction with IFPMA and the newly formed Industry Coordinating Group on Rare Diseases aims to ensure the momentum from the historic UN Resolution and needs of PLWRD are featured in the UHC debate within the private sector.

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, during 2022 RDI has been exploring further the 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 2022, RDI organized two internal strategic meetings to discuss the GSoAR project and how it interacts with several work streams and priorities within the overall RDI Strategy and annual Action Plan. Also, RDI and its partners (EURORDIS and Orphanet) needed to get legal advice for the establishment of a Consortium Agreement to govern their collaboration in relation to the GSoAR. The GSoAR would constitute a Rare Disease policy information resource dedicated to providing State of the Art data on rare disease policies and stakeholders’ landscape with the aim to progressively establish a global reliable information resource on the rare disease policy landscape. The ambition is for the GSoAR in Rare Disease to represent an independent and credible global resource that will be established by pooling data in collaboration between patient organisations, academia, clinicians, and national authorities and destined to be a factual, qualitative, and useful resource to different targeted stakeholders.
Acknowledgements

RDI would like to thank the following organizations, foundations, and companies for their financial support in 2022

RDI Members

A special mention to EURORDIS for funding and operational support

Foundations

Non-Health Companies

RDI Alliance of Companies
RDI Strategic Engagement Programme (formerly Capacity Building Programme)

Global Rare Disease Network

Global State of the Art Resource
Annex 1

Full List of RDI Member organizations as of December 2022

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 Seltener Erkrankungen 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 Outras Doencas 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. Coalition of Rare Diseases in Israel
23. Cutis Laxa Internationale
24. Cyprus Alliance For Rare Disorders
25. Debra International
26. EAT – Esophageal Atresia Global support groups
27. Esperanzaa
28. EURORDIS – Rare Diseases Europe
29. Fabry International Network
30. FEDER - Federación Española De Enfermedades Raras
31. Federación Argentina de Enfermedades Poco Frecuentes
32. Federacion Colombiana De Enfermedades Raras
33. Federación Mexicana de Enfermedades Raras
34. Federation of European Associations of Patients affected by Renal Diseases
35. Fondation Internationale Tierno et Miriam - Burkina Faso and Guinea
36. Fragile X International
37. Genetic Alliance Australia
38. Genetic Support Network of Victoria
39. Georgian Foundation for Genetic and Rare Diseases
40. Global Alliance of Sickle Cell Disease Organisations
41. Hypopara Norge
42. Illness Challenge Foundation
43. Indian Organization for Rare Diseases
44. Instituto Vidas Raras
45. International Alliance of Dermatology Patient Organizations
46. International Federation for Spina Bifida and Hydrocephalus
47. International Federation of Psoriasis Associations
48. International Gaucher Alliance
49. International Niemann-Pick Disease Alliance
50. International Patient Organization for Primary Immunodeficiencies
51. International Prader-Willi Syndrome Organisation
52. Japan Patient Association
53. Leukemia Patient Advocates Foundation
54. Malaysian Rare Disorders Society
55. MCT8-AHDS Foundation Inc.
56. Naevus Global
57. National Alliance for Rare Diseases Support - Malta
58. National Organization for Rare Diseases of Serbia
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<td>Nicolaides-Baraitser Syndrome Worldwide Foundation</td>
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<td>Organization For Rare Diseases India</td>
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<td>Pancyprian Federation of Patients’ Associations and Friends</td>
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<td>Pespa (Greek Alliance for Rare Diseases)</td>
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<td>PH Latin Society</td>
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<td>Philippine Society for Orphan Disorders</td>
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<td>Pro Rare Austria, Allianz für seltenen Erkrankungen</td>
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<td>Rare Disease Foundation of Iran</td>
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<td>Rare Diseases Sweden (Riksförbundet Sällsynta Diagnoser)</td>
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<td>World Federation of Hemophilia</td>
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