A strong common voice for Persons Living with a Rare Disease and their families around the world
2023 HIGHLIGHTS

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MESSAGE FROM THE COUNCIL CHAIR AND THE CEO

Message from the Chair and CEO

Dear Members and Supporters,

For Rare Diseases International (RDI), 2023 was a year marked by both transition and progress. As an organization, we began outlining a roadmap for the future and oversaw a change in leadership, with the appointment of a new Chief Executive Officer, two new Council members and three new team members. Thanks to the tireless efforts of our collective rare community, we were able to make significant advances in policy and practice.

We intensified advocacy efforts, calling for the inclusion of rare diseases in the United Nations Political Declaration on UHC. We participated in the review of the UN Resolution on Rare Diseases, successfully improving the text with stronger language on undiagnosed conditions and access to diagnosis. We contributed to the WHO Model List of Essential Medicines (EML) and took part in the Committee Open Forum, delivering an oral statement on behalf of the rare disease community. We led innovative collaborations to tackle fundamental challenges, including access to medicines and networked care models. We welcomed 7 new members, bringing our membership to 93 organizations in 48 countries. We also hosted an in-person membership meeting in Barcelona, bringing our members together and providing an opportunity for them to share essential insights on the future of the rare disease community. We invite you to celebrate these milestones with us and to reflect on how far the rare disease community has come.

Today, rare diseases are at a flex point. Once a disparate assortment of isolated conditions believed to affect only a few people, often in a devastating way, they were considered either too difficult to address or not worth diagnosing, treating or supporting. Now, science is advancing, healthcare and social service provisions are improving, and policies are emerging in more countries, translating into more people being diagnosed and living not only longer but better. This progress is the result not only of the work of researchers and clinicians but also the mobilization of patients and families.

RDI is proud to be part of this international patient-driven movement by supporting collaboration within the rare disease community, greater awareness of rare diseases, and global and regional advocacy. As the rare disease landscape evolves, RDI’s mission remains the same: to be a strong common voice on behalf of rare disease patients around the world, to advocate for rare diseases as an international public health priority and to represent its members and enhance their capacities.

As we look towards the future, RDI remains steadfast in advocating for equitable access to healthcare, promoting research, and raising awareness to ensure individuals with rare diseases receive the support and care they need. Despite the progress we have made, many challenges persist within the global health context: the lack of awareness and understanding, delays in diagnosis, misdiagnosis, limited access to appropriate treatments and support services, and disparities across the international landscape. There is an urgent need for healthcare systems to build on the experience gained over the last 20 years and to develop and implement innovative solutions for equitable and improved access to care and support for PLWRD. As continued advancements in research and technology offer more and better tailored solutions for rare diseases, RDI will ensure that the patient voice is
strong, insistent and united in ensuring transparency, ethical standards and patient rights.

We remain committed to addressing the systemic challenges that hinder access to healthcare treatments for individuals with rare diseases, particularly in under-served communities. We will continue to advocate for policy reforms, innovative health system financing solutions, and international cooperation to achieve greater equity in healthcare access.

In 2024, RDI will pursue several strategic priorities aimed at creating a lasting impact in the rare disease landscape. Foremost is our commitment to strengthening membership in our global alliance. By expanding our network of member organizations and fostering collaboration, we aim to amplify our collective voice and enhance our capacity to advocate for persons living with rare diseases worldwide. We are also committed to strengthening our regional alliances, recognizing their key role as an interface between global and local action.

A major advocacy objective for this year is the adoption of a comprehensive Global Action Plan for Rare Diseases at the World Health Assembly. This landmark initiative seeks to elevate the profile of rare diseases on the global health agenda, catalyze coordinated action across sectors, and drive tangible progress towards addressing the unmet needs of individuals with rare diseases.

RDI is also committed to forging strategic partnerships with global stakeholders, including governments, international organizations, academia, and industry. By harnessing the collective expertise, resources, and influence of these partners, we can leverage synergies, drive innovation, and unlock new opportunities to advance rare disease advocacy on a global scale.

We call upon our members, partners, and supporters to join us in this mission – to lend their voices, expertise, and resources to ensure that no one affected by a rare disease is left behind.

We extend our heartfelt gratitude to the rare disease community, our members, partners, and supporters for their unwavering commitment to improving outcomes for the over 300 million people living with a rare disease worldwide. Together, we have the power to shape a future where people living with rare diseases experience full access, equity and inclusion, no matter where in the world they are.

Sincerely,

ALEXANDRA HEUMBER PERRY
CEO

DURHANE WONG-RIEGER
COUNCIL CHAIR
WHO WE ARE

RDI is the global alliance of people living with a rare disease (PLWRD) across all countries and all rare diseases. We strive for a world where PLWRD and their families experience a better life through full recognition and support.

The membership of RDI brings together more than 90 organizations, representing the rare disease community in over 150 countries worldwide.

RDI enhances the capacities of an inclusive and diverse community of members and patient leaders across the globe. At international institutions, we represent an informed, independent and person-centered voice advocating for equity, Universal Health Coverage and improved outcomes for PLWRD. Through our global programmes, we foster and lead multi-stakeholder collaborations that aim to strengthen health systems for all people living with a rare disease.

HOW WE WORK

ADVOCATE

For rare diseases and PLWRD as a global policy priority

REPRESENT

PLWRD and their families at international institutions

SUPPORT

The empowerment of RDI members and patient advocate leaders
# OUR IMPACT STRATEGY

## Our Vision
A world where Persons Living with a Rare Disease (PLWRD) and their families experience a better life through full recognition and support.

## Our Mission
Be a strong common voices for PLWRD and their families around the world

## Our Values
Global  Equitable  Inclusive  Diverse  Person-centered  Independent  Collaborative  Innovative

## Improve EQUITY for PLWRD around the world

## Our Strategic Goals

<table>
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<tr>
<th>Community Engagement</th>
<th>Prioritization of rare disease in policy</th>
<th>Care pathways and networks</th>
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<tr>
<td>Enhance capacity and engagement of rare diseases community in the response</td>
<td>Financing for adequate and sustainable resources for rare disease care</td>
<td>Improve access to treatment and care for PLWRD</td>
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</tbody>
</table>

## Our Strategic Pathways

### STRATEGIC ENGAGEMENT
Building capacity fellowships, toolkits and resources, trainings, grants
Strengthening regional alliances regional events, dialogues and collaboration
Raising awareness Rare Disease Day, communication and awareness raising

### GLOBAL ADVOCACY
Shaping the global policy ecosystem
UN Resolution on PLWRD and their families
UN Political Declaration on UHC
Advocating for PLWRD as a global advocacy priority
WHA Resolution, Global Action Plan
Partnering on a global and regional scale
UHC2030, NGO Committee for Rare Diseases

### GLOBAL PROGRAMMES
Improving recognition of rare diseases
Operational Description of RD
Supporting timely diagnosis
Newborn screening, WHO EDL, Global Commission
Advocating for PLWRD as a global advocacy priority
WHA Resolution, Global Action Plan
Partnering on a global and regional scale
UHC2030, NGO Committee for Rare Diseases

### STRATEGIC PARTNERSHIPS
Fostering partnerships for impact
Foundations, Philanthropic organizations, public sector donors
Engaging the private sector
RDI Alliance of Companies
Sustainable resource development

## Our Strategic Enablers
Governance and Membership
Team and organizational culture
Operations and Infrastructure
Financial Sustainability and Efficiency
Throughout this period of transition and growth for RDI, our mission of improving the lives of persons living with rare diseases (PLWRD) at the global level remains at the core of our activities. We continue to strive to be a strong common voice on behalf of rare disease patients around the world and to represent our members and their priorities in a collaborative, inclusive way.

We are committed to promoting equity, advocating for policy reforms, and seeing innovative solutions to the systemic challenges facing people with rare diseases in terms of access to care. Our 2023 activities were founded upon these commitments and form the building blocks for our future actions.

**OUR IMPACT STRATEGY**

**ADVOCATING FOR RARE DISEASES AS AN INTERNATIONAL POLICY PRIORITY**

In 2023, our advocacy efforts focused on improving equity and access for the rare disease community. In September, we held a formal side event at the UN High-Level Meeting on Universal Health Coverage, calling on Member States to ensure that no one is left behind in UHC, notably by ensuring that rare diseases were included in the scope of the Political Declaration on UHC. The high-level, global participation in our side event, including 5 Ministers of Health, 4 other high-level representatives of state, 2 UN agencies, 3 international institutions and patient advocates from 5 countries, provided further evidence that the UN Member States are collectively committed to supporting the rare disease community with the objective of meeting their complex needs. It also underlined the progress that has been made in some countries, such as Malaysia and Colombia, through the mobilization led by RDI members.

We also participated in the review of the UN Resolution on Rare Diseases, successfully improving the text with stronger language on undiagnosed conditions and access to diagnosis. We contributed to the WHO Model List of Essential Medicines (EML) and took part in the Committee Open Forum, delivering an oral statement on behalf of the rare disease community.
LEADING THE WAY TOWARDS A GLOBAL ACTION PLAN ON RARE DISEASES

In 2024, we are seeking to build on this momentum to achieve long-lasting improvements for the rare disease community through a World Health Assembly Resolution calling for a Global Action Plan on Rare Diseases. This initiative seeks to transform the commitments made by Member States into tangible progress by establishing guiding principles, setting clear targets and deadlines, enabling accountability, and providing a dedicated budget for implementation.

IMPROVING ACCESS THROUGH INNOVATIVE COLLABORATION

In 2023 we completed the impact assessment on the Operational Description of Rare Diseases and released the “what is a rare disease” video. We gathered perspectives from 55 global stakeholders from 47 organizations with varied roles across research and development, regulatory and funding agencies, and healthcare service settings. The reference document focuses on three areas: building a common understanding of rare disease, improving the recognition and visibility of PLWRD, and catalyzing action.

We worked with international partners to find innovative ways to address fundamental challenges, including access to medicines and networked care models. We coordinated the IRDiRC-RDI Access Working Group and initiated research into barriers to accessing rare disease medicines, especially in low- and middle-income countries. Two case studies were completed, with input from patient representatives and clinicians from 17 countries. The learnings from these studies will support future development of an access framework to promote changes in a systemic way. For the first time, RDI was present and delivered an oral statement during the Open Session of the 24th WHO Expert Committee on the Selection and Use of Essential Medicines. We underlined importance for the Committee to apply appropriate selection criteria for rare disease medicines and making those medicines more visible to policy makers.

We continued our collaboration with the rare disease community and WHO to advance the Global Network for Rare Diseases (GNRD) project. Through both community building and knowledge exchange, the GNRD will address unmet needs for PLWRD, providing possibilities for capacity building and advance the quality of care, as well as strengthening health systems. In 2023, the Panel of Experts, composed of over 200 experts from across the world who contributed to shaping the project’s framework, transformed into the Multi-stakeholder Forum. This forum now serves as a platform for open dialogue and driving bottom-up approaches. We delivered the updated Operational Framework and Annexes to the WHO, and organized the interview series #GN4RARE to showcasing best practices from experts. Continuing project implementation, a technical global workshop was organized by RDI in December to develop recommendations on the final technical research necessary to set up the proof-of-concept network.

BUILDING CAPACITY AND CONNECTIONS THROUGH MEMBER ENGAGEMENT

We hosted an in-person membership meeting in Barcelona, bring our members together and providing an opportunity for them to share essential insights on the future of the rare disease community. We enhanced our commitment to building and enhancing regional alliances by supporting and participating in member-led meetings in Latin America and Asia, and strengthened local and national capacity by providing fellowships for our members to attend international events such as the World Orphan Drug Congress-USA, the World Orphan Drug Congress – Europe, and the UN High-Level Meeting on UHC at the UN Headquarters in New York City.

We expanded our membership by adding 7 new members, including three new national alliances and four new disease-specific organizations.

IMPACT HIGHLIGHTS

- 102 RDI MEMBERS AS OF APRIL 2024
- 25 FELLOWSHIPS AWARDED TO PATIENT LEADERS FROM 20 COUNTRIES
- 2 UN HIGH-LEVEL DOCUMENTS PRIORITISE RARE DISEASES
- 220 EXPERTS ENGAGED IN GLOBAL PROGRAMMES
# A Year in Review

<table>
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<tr>
<th>JANUARY</th>
<th>FEBRUARY</th>
<th>MARCH</th>
<th>APRIL</th>
<th>MAY</th>
<th>JUNE</th>
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<tbody>
<tr>
<td>Launch of the “What is a Rare Disease” animated video that gives a clear and inclusive definition of rare diseases.</td>
<td>#SEERARE, the Rare Disease Day event is viewed over 1200 times on Facebook. 32 countries submit video messages.</td>
<td>10th Alliance of Companies Meeting focuses on global rare disease advocacy.</td>
<td>The Chair of the RDI Council addresses the 24th WHO Expert Committee on the Selection and Use of Essential Medicines.</td>
<td>RDI fellows from Kenya and Peru attend the World Orphan Drug Congress (WODC) USA in Washington.</td>
<td>The Access Working Group completes 2 case studies on cystinosis and cystic fibrosis. The studies covered 11 and 7 countries respectively.</td>
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<tr>
<td>RDI presents a session on “systems thinking towards access” at the IRDiRC Conference – RE(ACT) Congress.</td>
<td>RDI partners with ALIBER for the High Level Forum - X Iberoamerican Meeting on Rare Diseases in Costa Rica.</td>
<td>The Thai Rare Disease Foundation joins RDI.</td>
<td>RDI and GARDaccess are joined by patient advocate groups at WODC USA for a panel discussion on access in LMICs.</td>
<td>Annual General Meeting takes place on-line. Two new Council members are elected: Kirsten Johnson, (Fragile X International) and Monica Ferrie (Genetic Support Network of Victoria).</td>
<td>Alexandra Heumber Perry takes the helm as RDI’s new CEO.</td>
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## A YEAR IN REVIEW

<table>
<thead>
<tr>
<th>JULY</th>
<th>AUGUST</th>
<th>SEPTEMBER</th>
<th>OCTOBER</th>
<th>NOVEMBER</th>
<th>DECEMBER</th>
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<tr>
<td>The updated WHO Model List of Essential Medicines is published and the Expert Committee’s conclusions include rare diseases.</td>
<td>The 11th Alliance of Companies Meeting delves into the Global Network for Rare Diseases.</td>
<td>RDI organized a momentous side event at the UN High-level Meeting (HLM) on Universal Health Coverage.</td>
<td>We host a closed Membership Event to discuss the future of the global rare disease alliance.</td>
<td>The regional conference programme supports the APARDO Conference in Malaysia. The event brings together over 150 stakeholders.</td>
<td>The WHO Secretariat determines that RDI meets the eligibility criteria for accreditation as a Non-State Actor in Official Relations Status to the WHO. The application will be presented in 2024.</td>
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### JULY 2023 Highlights

20 RDI fellows from 20 countries attend the Membership Event and alongside the WODC, Europe in Barcelona.

### AUGUST 2023 Highlights

The UN Resolution on PLWRD and their families is reviewed and contains stronger language on rare disease diagnosis and the role of civil society and the WHO.

### SEPTEMBER 2023 Highlights

Member States recommit to Political Declaration on UHC at the UN HLM. The text pinpoints rare diseases as a priority area.

### OCTOBER 2023 Highlights

The Global Network for Rare Diseases holds a two-day technical workshop in Singapore. The event convenes the WHO technical officer and rare experts.

### NOVEMBER 2023 Highlights

Huntington Association, RD Society of Nepal, Skraban-Deardorff Syndrome Foundation, United Porphyrias Association, Centre Alliance (Rwanda), Casa Hunter (Brazil) join RDI.
MEMBERS ACROSS THE GLOBE

Through RDI, patient-driven organizations come together to advance equity for PLWRD worldwide. Our diverse membership forms a robust global network, empowering patient groups, shaping international policy, and leading regional and global initiatives to improve outcomes for PLWRD.

Bring together organizations or patient advocates from a wide range of rare diseases in one country

**NATIONAL ALLIANCES**
- EURORDIS (Europe)
- ALIBER (Iberoamerica)
- ERCAL (the Caribbean and Latin America)
- APARDO (Asia-Pacific)

Bring together alliances for the same rare disease or disease area at the international or regional level.

**DISEASE-SPECIFIC FEDERATIONS AND GLOBAL PLATFORMS**
- 51 NATIONAL ALLIANCES
- 47 DISEASE-SPECIFIC FEDERATIONS AND GLOBAL PLATFORMS

NEW NATIONAL ALLIANCES
- Europe (EURORDIS), Asia-Pacific (APARDO), Iberoamerica (ALIBER) and the Caribbean and Latin America (ERCAL)

RDI is actively facilitating the creation of an African rare disease alliance.

NEW MEMBERS AS OF APRIL 2024
- Casa Hunter (Brazil), Federación Ecuatoriana de Enfermedades Raras (Ecuador), Instituto Unidos Pela Vida (Brazil), Interessengemeinschaft Hämosthen (Huntington's Disease), International Huntington Association, International Pemphigus and Pemphigoid Foundation, Rare Diseases Society of Nepal, Snelken-Deardorff Syndrome Foundation, Tewawuma Assistance Handicap (Senegal), TESS Research Foundation, The Rare Disease Foundation, Undiagnosed Diseases Network Foundation, United Porphyrias Association, Centre Alliance (Rwanda)
## RDI Members

Full alphabetical list as of April 2024

<table>
<thead>
<tr>
<th>Organization Name</th>
<th>International Organization Name</th>
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<tbody>
<tr>
<td>Advocacy Service for Rare and Intractable Diseases (Japan)</td>
<td>Interessengemeinschaft Hämophiler e.V.</td>
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<td>Ali Kimara Rare Disease Foundation</td>
<td>International Alliance of Dermatology Patient Organizations</td>
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<td>Alianza Argentina de Pacientes (Argentina)</td>
<td>International Federation for Spina Bifida and Hydrocephalus</td>
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<td>Alliber-Alianza Iberoamericana de Enfermedades Raras</td>
<td>International Federation of Psoriasis Associations</td>
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<td>Alliance Maladies Rares (France)</td>
<td>International Gaucher Alliance</td>
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<td>Alliano Chronicer Seltener Erkrankungen e.V. (Germany)</td>
<td>International Huntington Association</td>
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<td>Arabic Organisation for Rare Diseases</td>
<td>International Niemann-Pick Disease Alliance</td>
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<td>Asia Pacific Alliance of Rare Disease Organisations</td>
<td>International Patient Organization for Primary Immunodeficiencies</td>
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<td>Asociación de Familiares y Afectedos por Lipodistrofias</td>
<td>International Pemphigus and Pemphigoid Foundation</td>
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<td>Associacao Brasileira de Enfermedades Raras (Brazil)</td>
<td>International Prader-Willi Syndrome Organisation</td>
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<td>Associacao Portuguesa CDG e Outras Doencas Metabolicas</td>
<td>Japan Patient Association</td>
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<td>Association Anna (France)</td>
<td>Leukemia Patient Advocates Foundation</td>
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<tr>
<td>Association Aux Pas du Coeur (Cote d’Ivoire)</td>
<td>Malaysian Rare Disorders Society</td>
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<td>Beacon for Rare Diseases (United Kingdom)</td>
<td>MCT8-AHDS Foundation Inc.</td>
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<td>Blackswan Foundation</td>
<td>Naevus Global</td>
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<td>Botswana Organization for Rare Diseases</td>
<td>National Alliance for Rare Diseases Support – Malta</td>
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<td>Canadian Organization for Rare Disorders</td>
<td>National Organization for Rare Diseases of Serbia</td>
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<td>Casa Hunter (Brazil)</td>
<td>Nicolaides-Baraitser Syndrome Worldwide Foundation</td>
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<td>CDH International</td>
<td>Nord National Organization for Rare Disorders – NORD (USA)</td>
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<tr>
<td>Centre Alliance (Rwanda)</td>
<td>Organization For Rare Diseases India</td>
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<tr>
<td>Child &amp; Youth Care, Zimbabwe</td>
<td>Pancyprian Federation of Patients’ Associations and Friends</td>
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<td>China Alliance For Rare Diseases</td>
<td>Pespa (Greek Alliance for Rare Diseases Network)</td>
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<td>Chinese Organization for Rare Disorders</td>
<td>PH Latin Society</td>
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<tr>
<td>CMTC-OVM</td>
<td>Philippine Society for Orphan Disorders</td>
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<tr>
<td>Coalition of Rare Diseases in Israel</td>
<td>Pro Rare Austria (Allianz für seltenen Erkrankungen)</td>
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<tr>
<td>Cutis Laxa Internationale</td>
<td>Rare Disease Foundation of Iran</td>
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<td>Cyprus Alliance For Rare Disorders</td>
<td>Rare Disease Hong Kong</td>
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<td>Debra International</td>
<td>Rare Diseases Croatia</td>
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<td>Disease Organisations Hyppora Norge</td>
<td>Rare Diseases Ghana Initiative</td>
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<tr>
<td>EAT – Esophageal Atresia Global support groups</td>
<td>Rare Diseases Lesotho Association</td>
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<tr>
<td>Ehlers-Danlos Society</td>
<td>Rare Diseases Society of Nepal</td>
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<tr>
<td>Enfermedades Raras en el Caribe y América Latina (ERCAL) (the Caribbean and Latin America)</td>
<td>Rare Diseases South Africa</td>
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<tr>
<td>Esperanza (Peru)</td>
<td>Rare Diseases Sweden (Riksförbundet Sällsynta Diagnoser)</td>
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<tr>
<td>EUORDIS – Rare Diseases Europe</td>
<td>Rare Disorders Kenya</td>
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<tr>
<td>Fabry International</td>
<td>Rare Disorders NZ (New Zealand)</td>
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<td>FEDER – Federación Española De Enfermedades Raras (Spain)</td>
<td>Rare Voices Australia</td>
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<td>Federación Argentina de Enfermedades Poco Frecuentes (Argentina)</td>
<td>Retina International</td>
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<td>Federacion Colombiana De Enfermedades Raras (Colombia)</td>
<td>Romanian National Alliance for Rare Diseases</td>
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<td>Federación Ecuatoriana de Enfermedades Raras (Ecuador)</td>
<td>Russian Patient Association</td>
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<tr>
<td>Federación Mexicana de Enfermedades Raras (Mexico)</td>
<td>Skraban-Deardorff Syndrome Foundation</td>
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<td>Federation of European Associations of Patients affected by Renal Diseases</td>
<td>Taxawuma Assistance Handicap (Senegal)</td>
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<td>Fondation Internationale Tierno et Miriam (Burkina Faso and Guinea)</td>
<td>TESS Research Foundation</td>
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<td>Fragile X International</td>
<td>Thai Rare Disease Foundation</td>
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<td>Genetic Alliance Australia</td>
<td>Thalassaemia International Federation</td>
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<td>Genetic Support Network of Victoria (Australia)</td>
<td>Timothy Syndrome Alliance</td>
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<td>Georgian Foundation for Genetic and Rare Diseases</td>
<td>Undiagnosed Diseases Network Foundation</td>
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<td>Global Albinism Alliance</td>
<td>Unidos Pela Vida</td>
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<td>Global Alliance of Sickle Cell</td>
<td>United Porphyrias Association</td>
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<td>Illness Challenge Foundation (China)</td>
<td>Wilhelm Foundation</td>
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<tr>
<td>Indian Organization for Rare Diseases</td>
<td>World Alliance of Pituitary Organizations</td>
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<tr>
<td>Instituto Vidas Raras (Brazil)</td>
<td>World Duchenne Organization</td>
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BUILDING AND GROWING A DIVERSE GLOBAL ALLIANCE

In 2023, RDI welcomed seven new members, growing its membership to 93 groups registered in 43 countries and actively engaged in over 150 countries. We warmly welcome the following new members: the International Huntington Association, the Thai Rare Disease Foundation, the Rare Diseases Society of Nepal, the Skraban-Deardorff Syndrome Foundation, the United Porphyrias Association, Centre Alliance (Rwanda), and Casa Hunter (Brazil).

“This is an opportunity to collaborate globally, share expertise and research and support rare diseases in Thailand. Being part of RDI strengthens our efforts in raising awareness, advocating for rare diseases and accessing resources and knowledge from the international community”.

Pawerisa Assawasoontonnet
Thai Rare Disease Foundation
The 9th Membership Meeting was held online on 15 June. The annual gathering of all our members follows the Annual General Assembly and serves as an essential moment for consultation and engagement on RDI’s strategy, priorities and activities.

Speakers and attendees reflected on strategies to enhance the capacity of patient groups, the impact of international policy on national health systems, and the evolution of global collaborations and multi-stakeholder partnerships.

The agenda included perspectives and lessons learnt from national alliances in Ghana, Kenya, Malaysia, New Zealand, Serbia, the USA, and Zimbabwe, as well as contributions from the International Gaucher Alliance, the Asia Pacific Alliance of Rare Disease Organisations (APARDO) and the Iberoamerican Alliance for Rare Diseases (ALIBER).

**TAKE-HOME MESSAGES**

1. **Foster regional rare diseases alliances** to address specific regional and national challenges
2. **Leverage the UN Resolution on PLWRD** as a framework for action to guide policymakers and UN Agencies.
3. **Nurture a network of experts** and continue to work with WHO to promote the development of centres of excellence for rare diseases.
4. **Promote holistic approaches to care** for PLWRD and their families.
In the long-awaited first in-person meeting since the COVID-19 Pandemic, 40 members from 6 continents gathered in Barcelona, Spain, to share their visions, hopes and goals for the future of PLWRD in their regions and across the globe.

The room was buzzing with excitement as participants were encouraged to share ideas and think creatively to help chart the next chapter for rare disease advocacy and identify opportunities for collaborative global action.

Acknowledging the unique challenges faced by PLWRD in each country and region, the group identified key common themes critical to the entire community: equity, inclusion, access, Universal Health Coverage and representation.

These shared priorities will play a pivotal role in shaping and guiding the vision and strategy of RDI in the years to come.
Following the conclusion of the closed session, the group was joined by several partners from RDI’s Alliance of Companies for a **multi-stakeholder panel discussion**. Moderated by RDI Chair Durhane Wong-Rieger, the panel featured Antoine Gliksohn (Global Albinism Alliance), Mirjam Mann (ACHSE - Germany), Eda Selabatso (BORDIS - Botswana), Monica Ferrie (Genetic Support Network of Victoria - Australia), Regina Prospero (Instituto Vidas Raras - Brazil), and Jiaying Zheng (China Alliance for Rare Diseases).

The panellists shared inspiring stories of achievements and challenges they faced while advocating for rare diseases in their country or specific rare disease area. They highlighted the value and potential of a global alliance coordinating joint action to promote equity and improve outcomes for PLWRD.

"There are many commonalities among the RDI Members. There is no doubt that by joining forces, much progress can be made in addressing the many challenges faced by the rare disease community around the world"

**Antoine Gliksohn** (far right in image above)

**Global Albinism Alliance**
The Strategic Engagement Programme is a collaborative effort with RDI Members, designed to support the membership community and ensure their meaningful engagement throughout RDI’s activities.

The Programme designs and disseminates resources and toolkits to strengthen rare disease advocacy, facilitates networking and knowledge sharing through event fellowships and regional conference support, oversees RDI engagement in the international Rare Disease Day campaign, and coordinates community participation in committees and international working groups.

**SHARING KNOWLEDGE - ENGAGE & CONNECT**

Engage & Connect is a micro-site enabling patient advocates to access multilingual resources and tools. The site promotes open fellowship applications, provides digital maps highlighting rare disease policies worldwide, and includes an event calendar showcasing regional conferences and international events. Additionally, visitors can find recordings of small group meetings hosted by RDI members, covering various relevant topics, including developing patient registries and advocating for national plans.

In 2024 the programme will enhance its materials on UHC and the Operational Description of Rare Diseases and develop a new toolkit to support the community’s call for WHA Resolution and a Global Action Plan.
SUPPORTING REGIONAL ALLIANCES - REGIONAL CONFERENCES

Regional Conferences, organized by members in partnership with RDI, serve as key platforms to foster dialogue and nurture collaborations and initiatives to address specific regional challenges and opportunities. In collaboration with its regional alliance members, RDI identifies international speakers and provides financial support, which enables the participation of patient advocates and supports event logistics.

In 2023, RDI partnered with Asia Pacific Alliance of Rare Disease Organisations (APARDO) and Iberoamerican Alliance for Rare Diseases (ALIBER) and continued its engagement with rare disease groups in Africa to foster the development of an African alliance.

2023 REGIONAL CONFERENCES IN NUMBERS

2 REGIONAL CONFERENCES
- Asia-Pacific (APARDO)
- Iberoamerica (ALIBER)

1700 VIRTUAL ATTENDEES

150 STAKEHOLDERS

At ALIBER’s annual meeting, 120 attended the meeting in person in Costa Rica in February.

Shared perspectives at APARDO’s conference in Malaysia. Attendees from 15 countries joined.
The Iberoamerican region convened on 9 – 10 February in Costa-Rica for an interactive two-day hybrid meeting. The gathering brought together 120 in person attendees while over 1700 connected virtually. Participants included rare disease groups, ministerial representatives from 11 countries, clinicians, regulators, and PLWRD.

Thematic discussions delved into regional challenges in the implementation of the UN Resolution on PLWRD, the role of centres of expertise, and best practices in social and health care. Participants actively contributed to a consensus document aimed at significantly improving outcomes for the over 40 million PLWRD in Latin America.

RDI Chair, Durhane Wong-Rieger, highlighted the importance of creating spaces for learning and knowledge-sharing. As an official partner of the event, RDI provided funding, supported the participation of patient advocates, and contributed to the agenda, reinforcing its commitment to fostering regional solutions through dialogue and collaboration.

"The High-Level Forum is an opportunity for speakers and the public to share experiences and jointly construct conclusions and statements. The consensus document will serve as a roadmap to comprehensively address the care of people living with a rare disease in the region".

Alicia María Males Henao
ALIBER
APARDO CONFERENCE 2023

Co-hosted by Asia Pacific Alliance of Rare Disease Organisations (APARDO) and the Malaysian Rare Disorders Society (MRDS), the APARDO Conference took place on 24 – 26 November in Kuala Lumpur. It convened 150 stakeholders from 15 countries across the Asia-Pacific region, including Australia, China, Indonesia, India, Korea, Malaysia, Nepal and the Philippines.

"The conference was centred on the patient’s journey, fostering collaboration, raising awareness, and advancing the collective understanding of rare disease".

Monica Ferrie
APARDO

RDI CEO Alexandra Heumber Perry, along with four RDI Council Members - Durhane Wong-Rieger, Ritu Jain, Monica Ferrie, and Rachel Yang - presented our 2024 action plan and reflected on how global and regional actions complement each other to enhance ecosystems for PLWRD. Our team highlighted the urgent need for a global framework for action to translate commitments made at the international level into action at the regional, national, and local levels.

In a significant and uplifting message of support, the Malaysian Ministry of Health pledged to prioritize PLWRD in its health agenda, ensuring that no one is left behind. This commitment includes the establishment of a dedicated fund for rare diseases and the endorsement of calls for a global framework for rare diseases.

The event highlighted the exciting potential for regional collaborations to raise awareness and inspire action from national stakeholders and public policymakers.

"It was important to us that PLWRD and patient groups from across the region sit at the table, discussing and ideating strategies with other stakeholder groups. We believe discussions are incomplete without PLWRD. We are not case studies. We are core stakeholders".

Nadiah Hanim Abdul Latif (far right in image above)
Malaysian Rare Disorders Society
Fostering Community Leaders – RDI Fellowships

Fellowships offer essential support for RDI members by enabling patient advocates to participate in workshops, meetings, and conferences worldwide. Through its fellowship programme, RDI helps foster a robust support network within the patient advocate community. At international and regional events, emerging leaders of the rare disease community establish meaningful connections with other stakeholders and enhance their organizational capacity.

In 2023, Christine Mutena from Rare Disease Disorders Kenya and Miriam Ramírez Lopez from the Association of Urea and Metabolic Cycle Disorders Peru received fellowships to attend the World Orphan Drug Congress (WODC) USA in Washington from 23 to 25 May.

"WODC, USA was an eye-opener for me as I was exposed to the world of industry. It was great for networking and opened new avenues for us to engage Clinical Research Organizations to benefit our members.

On a personal level, I also got to meet a clinician presenting on PIK3CA who really validated my concerns as a mother of a child with a rare disease".

Christine Mutena (right in image above)
Rare Disorders Kenya

23 more RDI fellowships were also awarded to members from 20 different countries on 6 continents.

In addition to participating in RDI’s in-person membership event on 29 October, the fellows were able to attend the World Orphan Drug Congress (WODC) Europe on 30 October to 2 November in Barcelona, Spain.

RDI Fellows represented the rare disease community in various capacities, including panel discussions, round-tables, and presentations. Their contributions covered a wide range of topics, including rare disease research, regulatory frameworks, and global advocacy, highlighting the extensive expertise within the rare disease community and providing a critical patient perspective at the event.
Rare Disease Day is an annual awareness-raising campaign which takes place on the last day of February and is led by national rare disease civil society groups worldwide.

As a coordinating partner, RDI promotes the participation of the global rare disease community outside the EU and Northern America and leverages the campaign to help raise awareness and advance common policy messages.

#SeeRare, RDI’s 2023 Rare Disease Day campaign, was co-organized with the Rare Disease Day campaign team and featured 32 video messages from RDI Members. The event aimed to spotlight the diverse rare community and tenacious network of patient-led organizations.

Each participant highlighted an aspect of life with a rare disease in their country and called for increased recognition and support for the over 300 million PLWRD. Hosted by NORD’s Lisa Sarfaty and KP Tsang from Rare Disease Hong Kong, the lineup showcased patient organizations from 32 countries, including Guinea, Argentina and Australia.

We wish to acknowledge our 2023 Strategic Engagement Programme Partners:
**GLOBAL ADVOCACY**

RDI’s global network plays a crucial role in shaping the global policy agenda, advocating for improved outcomes for people living with rare diseases (PLWRD) worldwide. Our community is dedicated to advancing transformative global policies, and supporting civil society groups to demand change and meaningful action on rare diseases at regional, national, and local levels.

In 2023, global rare disease advocacy achieved significant milestones as the UN and its agencies increasingly recognized PLWRD as a vulnerable group, emphasizing the importance of integrating their needs across the UN agenda. Moreover, at international events and regional meetings throughout the year, Member States and national ministries publicly committed to establishing national frameworks and implementing measures to address rare diseases.

This progress signifies a growing momentum toward equity for our community. RDI and its members are poised to drive advocacy efforts, ensuring that political statements translate into tangible action.
GROWING MOMENTUM AT THE UN - UN HIGH-LEVEL MEETING ON UHC

On 21 September, RDI partnered with EURORDIS – Rare Diseases Europe, Ågrenska, Federación Española de Enfermedades Raras (FEDER) and the NGO Committee for Rare Diseases to host a momentous side event at the UN High-Level Meeting on UHC in New York. The event, titled “Engaging the UN System & Member States to Achieve Universal Health Coverage for Persons Living with a Rare Disease: A Blueprint for Leaving No One Behind,” provided a powerful platform to emphasize the critical importance of including the rare disease community in Universal Health Coverage (UHC).

Co-sponsored by Spain, Sweden, and Qatar, the gathering was opened by Her Majesty Queen Letizia of Spain and Her Majesty Queen Silvia of Sweden. Queen Letizia highlighted the need for a WHA Resolution to mobilize Member States and pave the way toward equity for PLWRD. Queen Silvia underscored the significance of research and knowledge sharing in rare diseases.

The event garnered high-level support from national and regional policymakers, including the Ministers of Health of Spain, Sweden, Qatar, France, and Malaysia, as well as representatives from the Ministries of Health of Canada and Brazil, the Ministry of Foreign Affairs of the United Arab Emirates, and the European Commission. Discussions focused on the importance of a global approach, emphasizing the sharing of expertise, investment in scientific research, and alleviation of the economic burden of rare diseases.
Strong support from WHO, the World Bank, the Office of the UN High Commissioner for Human Rights (OHCHR), and the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO) reflects increasing political momentum since the 2019 Political Declaration on UHC, which included rare diseases for the first time, and the ground-breaking 2021 UN Resolution on PLWRD.

Patient advocates used this opportunity to recognise the significant progress that has been made and highlight ongoing challenges faced by those living with rare diseases, including delayed diagnoses, limited access to treatment, and financial strain.

RDI seized the invaluable moment to renew the global community’s push for a WHA Resolution on rare diseases, including a commitment to develop a Global Action Plan that provides a clear blueprint supported by funding and resources to ensure that UHC includes PLWRD and truly leaves no one behind.

**POLICY ACTION POINTS:**

1. Prioritise and integrate rare diseases within national UHC and Primary Health Care plans
2. Create and implement national plans or strategies for rare diseases involving PLWRD
3. Institute a specific funding stream for rare diseases
4. Strengthen primary health care and referral pathways to guarantee the early, timely and precise diagnosis of PLWRD
5. Address financial costs shouldered by PLWRD and their families
6. Include services which treat the complex needs of PLWRD and encompass a multidisciplinary and multi-sectoral approach
7. Develop national programs explicitly dedicated to rare diseases to enable rapid and equitable access to diagnosis and social support
8. Promote ethical and responsible international data sharing to support diagnosis, increase clinical collaboration, facilitate research and accelerate treatment of undiagnosed and rare conditions
9. Increase education and awareness of rare diseases in schools and public fora to dispel stigma and discrimination
10. Promote innovative solutions, including public-private partnerships for sharing knowledge and access to treatment
TOWARD TRULY UNIVERSAL HEALTH COVERAGE – RARE DISEASES INCLUDED IN UN DECLARATION ON UHC

On 21 September 2023, UN Member States gathered for the 2023 UN High-level Meeting (HLM) on universal health coverage (UHC). Member states recommitted to accelerating progress toward UHC by 2030, adopting a Political Declaration on UHC, which retains a mention of rare diseases, first included in the landmark 2019 Political Declaration on UHC.

The Political Declaration on UHC asserts that health is a fundamental human right and emphasises the importance of leaving no one behind in accessing health care and services. In the coming year, RDI will continue to advocate for greater recognition of rare diseases in the global UHC agenda. In 2024, we will amplify the call for a WHA Resolution, urging states to adopt national frameworks and make the necessary investments and actions to make UHC truly universal.

"It was so important for us to recognize the Declaration on UHC. This will give us the opportunity to work with our government, health ministries and clinicians to make the case for diagnosis, treatments and care".

Durhane Wong-Rieger (image above)
RDI Chair
IMPROVING THE UN RESOLUTION ON PLWRD AND THEIR FAMILIES

Participating through its Policy Committee, RDI took an active role in the consultation process to improve the groundbreaking UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families, a landmark document adopted in 2021. Led by the Permanent Mission of Spain to the United Nations in New York, this collaborative effort aimed to ensure that the resolution remains relevant and effective in addressing the needs of PLWRD.

Significant enhancements in the resolution’s text focus on key topics, including diagnostics, the pivotal role of civil society, and the engagement of the WHO in monitoring progress toward implementation.

THE AMENDED DOCUMENT INCLUDES:

1. New specific language on diagnosis, undiagnosed rare diseases and newborn screening
2. Calling on member states to strengthen “referral systems between primary health care and other levels of care”
3. Reference to the Political Declaration on UHC adopted by the UN in September 2023
4. A full paragraph inviting civil society groups AND NGOs to raise awareness of the challenges of PLWRD
5. An invitation to the Secretary-General, in close collaboration with the Director General of the WHO, to inform the General Assembly about the implementation of the present resolution during its 80th session in 2025
PARTNERING TO ACT ON A GLOBAL SCALE

Key partnerships with UN agencies, civil society platforms and international organizations bolster our community’s global advocacy.

In 2023, RDI maintained its active involvement as a member of the Civil Society Engagement Mechanism (CSEM) of UHC2030, a collaborative platform led by the WHO and World Bank to enhance health systems for UHC. As a UHC 2030 partner, resources and toolkits focusing on rare diseases and UHC are prominently featured on the CSEM Health for All Toolkit. Furthermore, through the platform, RDI can participate in global dialogues and campaigns to bolster health systems and expand UHC.

NGO COMMITTEE FOR RARE DISEASES

The NGO Committee for Rare Diseases, established Under the auspices of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO), is dedicated to fostering multi-stakeholder collaboration on rare diseases within the UN system. RDI partners with the Committee to engage the UN, including hosting the side event to the UN High-level Meeting.

WORLD HEALTH ORGANIZATION (WHO)

In December, the WHO Secretariat determined that RDI meets the eligibility and due diligence criteria for accreditation as a Non-State Actor in Official Relations Status to the WHO. RDI’s application will be presented to the Executive Board for consideration in 2024. While organizations often must apply several times to be granted this status, which is a privilege that the Board grants to nongovernmental organizations whose activities contribute significantly to the advancement of public health, this initial selection is an essential step towards achieving this important goal.

In addition, RDI has applied to join the WHO Civil Society Commission, which is tasked with strengthening dialogue and fostering collaboration between the WHO and civil society groups. The Commission also provides recommendations to support the WHO in effectively engaging civil society to achieve UHC and the Sustainable Development Goals.
Global Programmes harness the power of collaboration and knowledge-sharing across stakeholders to transform the lives of people with a rare disease.

RDI leads and partners in global initiatives advancing equity for PLWRD through increasing awareness amongst all stakeholders and promoting holistic approaches which positively impact the whole patient journey from diagnosis to access to expert care and social support. Under the umbrella of the Global Programmes are several key collaborations, including the Operational Description of Rare Diseases, as well as the Global Network for Rare Disease coordinated with the WHO and International Rare Diseases Research Consortium (IRDiRC).

**Along the Patient Journey**

**Access**
- WHO Model List of Essential Medicines (EML)
- Access Working Group
- GARDAccess
- International Rare Diseases Research Consortium (IRDiRC)

**Diagnosis**
- Newborn Screening
- WHO Model List of Essential In Vitro Diagnostics

**Recognition & Awareness**
- Operational Description of Rare Diseases

**Networked Care**
- Global Network for Rare Diseases (GNRD)
OPERATIONAL DESCRIPTION OF RARE DISEASES – IMPROVING RECOGNITION AND AWARENESS

In many countries, there needs to be more consensus on what constitutes a rare disease. Without a shared understanding of rare diseases, PLWRD remain invisible to most health systems and overlooked by social policies.

To enhance the recognition of rare diseases, RDI coordinated a global panel of experts (which included the WHO) to develop an Operational Description of Rare Diseases. This description not only facilitates the identification of rare diseases and those affected by them but also serves as a valuable tool for stakeholders seeking to improve their understanding of the unique challenges rare diseases present to health systems and society.

"Rare disease should not be purely defined by numbers. We should really understand what it means to live with a rare disease. The Operational Description delivers a useful and dynamic descriptive framework to tackle rare diseases".

Helen Zhou
China Illness Foundation

In January 2023, RDI hosted a webinar to launch “What is a Rare Disease,” an animated video that offers a clear and inclusive definition of rare diseases. The video’s engaging animated character and straightforward language make the Operational Description accessible to a broad audience, including PLWRD, patient advocates, healthcare professionals, and policymakers.

"Keeping varying definitions of rare diseases hinders their recognition and creates hurdles for healthcare access ".

Prasanna Shirol
ORDIndia

In March, the Impact Assessment Report on the Operational Description of Rare Disease was submitted to the WHO. The report notes that the Description is a valuable tool to build a common understanding of rare diseases. It also highlights that adopting the common description would be an important step to improving the recognition and visibility of PLWRD. Furthermore, the document will support further understanding of rare disease in the era of precision medicines.
SUPPORTING TIMELY AND ACCURATE DIAGNOSIS

Timely diagnosis and early access to care saves lives and reduces the impact of rare disease on the lives of patients and their support network. Despite advances in technology and innovation in science, PLWRD often face significant delays obtaining diagnosis, and are commonly misdiagnosed or undiagnosed.

In response RDI, engages in conversations and collaborations to support improved access to diagnosis and address disparities between regions and countries.

NEWBORN SCREENING

In February, RDI Programme Director, Mary Wang presented a global perspective on newborn screening (NBS), encouraging greater investments in early diagnosis at a workshop convened by Rare Diseases South Africa.

RDI also contributed as a co-editor to a special issue of the Rare Disease and Orphan Drugs Journal titled “Newborn Screening II - Policy, Ethics, and Patient Perspectives.” This special issue highlights obstacles to implementing national NBS programs and identifies opportunities presented by emerging technologies, such as next-generation sequencing.

To spark exchange on NBS as a public health priority, RDI arranged a panel session on patient perspectives on NBS at WODC, Europe in November.

WHO MODEL LIST OF ESSENTIAL IN VITRO DIAGNOSTICS

In March, RDI submitted the first analysis of the Essential Medicines List (EML) and the Essential in Vitro Diagnostics List (EDL) to the WHO, focusing on rare diseases. The EDL contains in vitro diagnostics which are useful for rare diseases. In addition, there was a call for submission for high priority tests including a number of rare diseases.

GLOBAL COMMISSION TO END THE DIAGNOSTIC ODYSSEY

RDI is a member of the Global Commission to End the Diagnostic Odyssey is a multi-disciplinary expert panel focused on fostering the exchange of ideas and launching innovative pilot projects to accelerate diagnoses and enhance equitable access to diagnostic services. Previous RDI initiatives supported by the Global Commission include developing a global survey of patient experiences of diagnosis. A new phase of pilot projects is scheduled to commence in 2024.
WORKING WITH WHO TO ADVANCE NETWORKED CARE
GLOBAL NETWORK FOR RARE DISEASES

The Global Network for Rare (GNRD) is driven by the vision of a world where all PLWRD, no matter where they live, have access to timely and accurate diagnosis and reliable quality care.

The GRDN is a pivotal component of RDI’s Memorandum of Understanding with the WHO. The proposed Network aims to pool existing expertise, encourage knowledge sharing, and facilitate virtual advice to ensure that PLWRD can access care through a global networked care model. The initiative aligns with WHO’s 13th Global Programme of Work, which seeks to expand UHC to include an additional 1 billion people.

In March 2023, the final annexe of RDI’s proposed operational framework for the GNRD was submitted to WHO, marking the culmination of the mapping and concept design phase of the project, and concluding the work of the Panel of Experts, a working group, coordinated by RDI and comprised of 221 experts from 75 countries.

The Panel played an essential role in outlining the global context for rare diseases, sharing best practices, and identifying opportunities for collaboration with existing networks. The group’s contributions have been instrumental in shaping the vision for GNRD and the operational framework. A series of interviews with members of the Global Network Panel of Experts #GN4RARE were published as a part of the community engagement campaign until January 2023. Papers include contributions on implementing national newborn screening programmes, twinning expert centres, cross-sector care models and digital health.

In 2024, RDI will consolidate the outcomes of the Panel discussions to develop resource maps highlighting rare disease best practices and collaborative care models worldwide.
To transition the GNRD from concept development to the proof-of-concept phase, RDI held a critical two-day technical workshop in Singapore on 14-15 December. The workshop convened the WHO focal point, Ulrike Schwerdtfeger, and seven clinical experts from diverse regions. Together, they reviewed the operational framework for the proposed GNRD and charted recommendations for additional technical research.

Clinical experts presented their existing services and models for providing care to PLWRD. They brought forth their diverse local realities and challenges, and suggested how GNRD can strengthen their activities. The workshop outlined key areas needing further technical research and solutions. These include: developing a sustainable financial model, establishing a legal framework that works across regions, and outlining a data management strategy to ensure information is secure and accessible.

The workshop’s findings will be compiled into concrete recommendations and presented to the WHO. This is a significant step towards making the GRDN a reality, ultimately improving access to diagnosis and expertise for all PLWRD.

"Gathering advocates from different parts of the world supports shared recommendations that apply to all. There is strength in collaboration".

Carmencita Padilla
University of the Philippines
Manila

Throughout the year, the Global Programme’s team maintained regular communication with the WHO to facilitate the initiative’s transition from the conceptual phase to a pilot phase coordinated by the WHO.

In addition, RDI also provided regular updates to stakeholders on the project’s progress, including presentations at the RDI Membership Meeting in June, and the 11th Alliance of Companies Meeting, a gathering that brings together our private sector donors.

"In 2019 WHO signed a Memorandum of Understanding with RDI to further our joint work. Since then, we have been working to create a Global Network for Rare Diseases, which will be a sustainable network of patient organizations, healthcare providers, researchers, and policymakers dedicated to rare diseases".

Jérôme Salomon,
Assistant Director-General
WHO
ACCELERATING ACCESS

Accessing standard-of-care treatments and cutting-edge therapies for rare diseases remains a significant challenge for PLWRD, particularly in low-and-middle-income countries (LMICs). Patients face a multitude of obstacles, including cumbersome regulatory pathways, prohibitive pricing, inadequate reimbursement, and a lack of health infrastructure for proper administration and monitoring of care.

RDI’s access initiatives leverage existing global access frameworks such as the WHO Model List of Essential Medicines (EML) and brings together multiple stakeholder groups to consider complex access barriers along the patient journey and pilot innovative solutions.

WHO MODEL LIST OF ESSENTIAL MEDICINES LIST

The inclusion of rare disease medicines in the WHO Model List of Essential Medicines (EML) and Essential Medicines List for Children (EMLc) is crucial for ensuring equitable access to treatment. RDI plays an active role in engaging with the WHO to advocate for integrating rare disease therapies in both the EML and the EML for Children.

RDI’s Council Chair, Durhane Wong-Rieger, addressed the 24th WHO Expert Committee on the Selection and Use of Essential Medicines on 24 April. Representing the global rare disease community, RDI urged Member States to accelerate efforts toward UHC and emphasized the importance of an inclusive approach when considering new entries to the EML to ensure persons with a rare disease have greater access to quality, affordable essential medicines and diagnostics.

KEY MESSAGES TO THE WHO EXPERT COMMITTEE

1. The inclusion of rare disease essential medicines in the WHO EML supports appropriate patient access in all countries
2. Appropriately apply the criteria and selection process to the realities of rare diseases and rare disease medicines
3. Improve annotation within EML to ensure medicines that could be relevant for rare diseases are clearly visible to users and policy makers
In 2021, RDI and the International Rare Diseases Research Consortium (IRDiRC) joined forces to launch the IRDiRC/RDI Global Access Working Group. This collaborative, multi-stakeholder initiative aims to comprehensively address barriers to access, with a particular focus on LMICs.

In February 2023, the Working Group completed two case studies. The first study examined cystinosis, involving interviews conducted in 11 countries. The second focused on cystic fibrosis and included data from 7 countries. These studies have provided valuable insights and lessons learned.

The learnings of the working group formed the basis of a dedicated session on “systems thinking towards access” at the IRDiRC Conference - RE(ACT) Congress in March as well as a session hosted at the WODC, USA in May 2023.

KEY TAKEAWAYS FROM WORKING GROUP CASE STUDIES:

1. **Adopt Holistic Approaches to Access**: Access to rare disease treatments involves the entire patient journey, from disease awareness to diagnosis, treatment, reimbursement, and ongoing care.

2. **Build Local Infrastructure**: There is a clear need for improved infrastructure at the local level to support better access to rare disease treatments.

3. **Raise Awareness and Improve Diagnosis**: Building awareness of rare diseases and establishing robust diagnostic infrastructure is a crucial initial step towards improving access for patients worldwide.

The learnings of the working group formed the basis of a dedicated session on “systems thinking towards access” at the IRDiRC Conference - RE(ACT) Congress in March as well as a session hosted at the WODC, USA in May 2023.

GARDACCESS

To build on the outputs of the Access Working Group, in 2023, RDI joined the GARDAccess initiative, led by the Partnership for Quality Medicines Donations (PQMD). GARDAccess is dedicated to accelerating patient access to medicines, treatments, and services. The initiative engages key stakeholders and is working to pilot innovative solutions that extend beyond traditional donation models.

Guidelines were developed in the first phase of the initiative in 2022 to outline the general principles to create partnership and alliances for PLWRD, focusing on improving the process for improved access to treatments.

In 2023 RDI continued to contribute its expertise and represent patient perspectives to further the initiative’s goals.
We continue to be actively support IRDiRC’s vision to enable all PLWRD to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention. IRDiRC is a global multi-stakeholder initiative aimed at fostering international research collaboration.

In 2023 our Council Chair Durhane Wong-Rieger continued to serve on IRDiRC's Operating Committee and Chair the Patient Advocate Constituent Committee. In addition RDI proposed and is leading a task force aimed as developing “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”.

We also contributed to the program design and took part in the RE(ACT) Congress and IRDiRC Conference in March. RDI and its members presented on topics from improving the recognition of rare disease to networked care models.

Supporting IRDIRC forms part of RDI’s strategic framework leveraging multiple stakeholders collaboration and encouraging cross-sectoral solutions that impact the lives of people with a rare disease.

“In the symphony of rare diseases, collaboration orchestrates the most beautiful harmony. We believe that by sharing knowledge and resources across borders, we can compose a global movement of care and support for people living with rare diseases and their families”.

Lisa Sarfaty
National Organization for Rare Disorders (NORD USA)
Nurturing strong partnerships across stakeholders is at the cornerstone of RDI’s strategy, enabling us to amplify our impact as a global community, advancing better outcomes for PLWRD worldwide.

By engaging a wide range of stakeholders, including UN agencies, international NGOs, philanthropic foundations, the private sector, and regional and global multi-stakeholder groups, we can leverage collective expertise, secure additional resources, and access a broader audience. With support from partnerships, we can access new insights, help drive innovative solutions, and improve our ability to advocate effectively on a regional and global scale.
ENGAGING THE PRIVATE SECTOR – RDI ALLIANCE OF COMPANIES

In 2023, RDI continued to strengthen its Alliance of Companies for Patient-Centered Action (AoC), a key platform for companies supporting the rare disease cause globally. The AoC is RDI’s primary channel for engaging with companies involved in rare diseases, offering them a unique opportunity to deepen their understanding of the international rare disease community and contribute to its success.

Biannual touch points on topics of mutual interest offer important moments for exchanging perspectives between the private sector and the rare disease community. In March, the AoC held its 10th Meeting, where discussions delved into global advocacy priorities. Members were also given the opportunity to provide feedback on the Key Asks of a proposed World Health Assembly (WHA) Resolution. The 11th AoC meeting in August shifted the focus to the Global Network for Rare Diseases, marking the project’s transitions from the concept development phase.

RDI ALLIANCE OF COMPANIES MEMBERS Alphabetical list as of December 2023

[Images of company logos]
OUR WORK AND THE COMMUNITY’S VOICE IS AMPLIFIED BY ENGAGING A WIDE-RANGE OF KEY STAKEHOLDERS AND WORKING IN COLLABORATION WITH OUR MEMBERS, UN ORGANIZATIONS, GOVERNMENTS, CIVIL SOCIETY GROUPS, FOUNDATIONS, ACADEMIC INSTITUTION, TREATMENT CENTRES AND THE PRIVATE SECTOR.

WE WOULD LIKE TO RECOGNISE THE FOLLOWING PARTNER ORGANIZATIONS IN 2023:

- Chan Zuckerberg Initiative
- CSEM
- Global Commission
- Fondation Philanthropia Europe
- IFPMA
- IRDiRC
- NGO Committee for Rare Diseases
- Telethon
- EURORDIS

WE WOULD ALSO LIKE TO ACKNOWLEDGE THE SUPPORT OF THE FOLLOWING UN AGENCIES AND BODIES:

- World Health Organization
- Office of the United Nations High Commissioner for Human Rights (OHCHR)
- Conference of NGOs in Consultative Relationship with the United Nations
RDI COUNCIL 2023

The RDI Council is our board of directors responsible for strategy, overview, resource sustainability and leadership. Individuals are elected by Full Member organizations at the Annual General Meeting (AGM) and serve for a three-year term. The Council is comprised of seven individuals, including the Council Chair, Treasurer, and Secretary.

At the AGM on 14 June 2023, two new members were elected to the Council: Kirsten Johnson, President of Fragile X International and Monica Ferrie, CEO of the Genetic Support Network of Victoria (GSNV) and a member of the Board of Directors of Asia Pacific Alliance of Rare Disease Organisations (APARDO).

DURHANE WONG-RIEGER
Chair
Canadian Organization for Rare Disorder (CORD)

MONICA FERRIE
Genetic Support Network Of Victoria (GSNV) and Asia Pacific Alliance of Rare Disease Organisations (APARDO)

YANN LE CAM
Treasurer
EURORDIS- Rare Diseases Europe

RITU JAIN
DEBRA International

RACHEL YOUNG
Secretary
China Alliance for Rare Diseases (CHARD) and Illness Challenge Foundation (ICF)

KIRSTEN JOHNSON
Fragile X International

KELLY DU PLESSIS
Rare Diseases South Africa
POLICY COMMITTEE

The Policy Committee recommends and develops RDI advocacy priorities, policies and key messages. In addition, the group co-creates RDI position papers, statements, and declarations. The Committee is composed of representatives from each WHO region as well as international federations.

YANN LE CAM  
Chair  
EURORDIS - Rare Diseases Europe

DAVID SANCHEZ  
Federacion Española de Enfermedades Raras (FEDER)

ELENI ANTONIOU  
Thalassaemia International (TIF)

LARA BLOOM  
Ehlers-Danlos Society (EDS)

ROBERTA DE PINA  
Federación Argentina de Enfermedades Poco Frecuentes (FADEPOF)

MIGDALIA DENIS  
Pulmonary Hypertension Latin Society

KELLY DU PLESSIS  
Rare Diseases South Africa

LUCIANA ESCATI  
Federación Argentina de Enfermedades Poco Frecuentes (FADEPOF)

ANTOINE GLIKSOHN  
Global Albinism Alliance

RITU JAIN  
Asia Pacific Alliance of Rare Disease Organisations

SALOME MEKHZULZA  
World Federation Hemophilia

CHRISTINE MUTENA  
Rare Disorders Kenya

RAMAIYAH MUTHYALA  
Indian Organization for Rare Diseases

JESUS NAVARRO  
Iberoamerican Alliance for Rare Diseases (ALIBER)

SYLVIA ROOZEN  
International Federation Spina Bifida and Hydrocephalus

LISA SAFARTY  
NORD - National Organization for Rare Disorders (USA)

LEIRE SOLIS  
International Patient Organisation for Primary Immunodeficiencies (IPOPI)

CHRISTINE WHITE  
Canadian Organization for Rare Disorders (CORD)
Under the leadership of our new CEO, Alexandra Heumber Perry, the RDI team is dedicated to realising the vision of a world where Persons Living with Rare Diseases receive full recognition and equity. Our diverse, dynamic, talented team works tirelessly to advance this mission, and we deeply value their unwavering commitment. Below is the staff list as of April 2024.

**RDI TEAM**

**ALEXANDRA HEUMBER PERRY**
Chief Executive Officer

**ALEXANDRA LIANES**
Executive Assistant

**MARCO PASETTI**
Finance and Administrative Manager

**DEBRA BELLON**
Strategic Engagement Manager

**NATACHA DEBBANÉ**
Senior Partnership Consultant

**HLAWULANI MKHABELA**
Strategic Communication Consultant

**HARSHINI KUMARI RATHORE**
Global Advocacy & Evidence Knowledge Intern

**MARY WANG**
Programme Director

**SARA BRAMBILLA**
Global Programme Junior Manager

**LUCIA MONACO**
Policy Advisor

**DANIEL WAINSTOCK**
Volunteer
DI’s 2023 financial accounts and report were prepared by RDI Finance & Administration Manager Marco Pasetti together with our Chartered Accountant Actheos, audited by Deloitte & Associates during March and April 2024.

Deloitte performed an ordinary audit of the financial statements (balance sheet, income statement and notes). The financial statements give Deloitte’s a true and fair view of the assets and liabilities and of the financial position of the Association as for December 31, 2023 and of the results of its operations for the year then ended in accordance with French accounting principles.

RDI total incomes in 2023 were 1.205 K€, with a decrease of 325 K€ compared to 2022, mostly due to the less In-Kind contributions received from both Eurordis and other volunteers. Total expenditures 936 k€, a decrease of 552 K€ on the previous year; mostly, is the balance effect of the leak of In Kind in revenues, but also some economies on staff and logistic fees.

In 2023 our incomes were diversified, with 48% from Health Corporates, 30% from Non-Health Companies, Foundations and other Non-For-Profit Organisations, 21% from from Patient Organisations.

Almost all assets of RDI, 663 K€ from a total of 698 K€ are liquid assets, cash on the accounts, composed principally by two liabilities: 341 K€ from the reserve and 269 K€ from the surplus of 2023.

2023 was a peculiar year for RDI: the 5-years Memorandum of Understanding 2019-2023 between EURORDIS and RDI, since its legal incorporation in 2018 until its planned full financial and administrative autonomy in 2024, has finished at the end of the year 2023. This MoU included the in-kind provision of a full time Executive director, the financial & HR & admin services, the office spaces. For this reason in 2023 RDI has, on one hand, continued to profit of Eurordis support, and on the other hand has started putting in place its new organisational structure. In June 2023, our new CEO, Alexandra Heumber-Perry has been appointed, with the aim to lead the Organisation. And a team has been formed over 2023-2024 to take over finance & administration and raise diversified funds.

As a results of all these transitions, RDI ended 2023 with a surplus of 269.463 €, enabling to propose to RDI members to increase its reserve by 80%, to a total of 610.254 €. This could assure about 6 months of functioning (based on 2024 budget) and let RDI continue to develop its advocacy, its programs, and the new organisational structure.
## BALANCE SHEET

<table>
<thead>
<tr>
<th>ASSETS</th>
<th>2023 EUR</th>
<th>2022 EUR</th>
<th>Δ</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intangible Fixed Assets</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Tangible Assets</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Trade notes &amp; accounts receivable</td>
<td>35 137</td>
<td>46 472</td>
<td>-11 335</td>
</tr>
<tr>
<td>Accruals</td>
<td>-</td>
<td>224</td>
<td>-224</td>
</tr>
<tr>
<td>Cash - Current Accounts</td>
<td>662 620</td>
<td>469 597</td>
<td>193 023</td>
</tr>
<tr>
<td><strong>TOTAL ASSETS</strong></td>
<td>697 757</td>
<td>516 292</td>
<td>181 465</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>LIABILITIES</th>
<th>2023 EUR</th>
<th>2022 EUR</th>
<th>Δ</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reserve</td>
<td>340 791</td>
<td>297 836</td>
<td>42 955</td>
</tr>
<tr>
<td>Profit/Loss for the period</td>
<td>269 463</td>
<td>42 955</td>
<td>226 508</td>
</tr>
<tr>
<td>Provisions for loss</td>
<td>432</td>
<td>2 951</td>
<td>-2 519</td>
</tr>
<tr>
<td>Accounts payables</td>
<td>48 058</td>
<td>33 445</td>
<td>14 613</td>
</tr>
<tr>
<td>Other Debts &amp; accruals liabilities</td>
<td>39 014</td>
<td>139 105</td>
<td>-100 091</td>
</tr>
<tr>
<td><strong>TOTAL LIABILITIES</strong></td>
<td>697 757</td>
<td>516 292</td>
<td>181 465</td>
</tr>
</tbody>
</table>
## Profit and Lost (With In-Kind Contributions)

<table>
<thead>
<tr>
<th>INCOME</th>
<th>2023 EUR</th>
<th>2022 EUR</th>
<th>Δ</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Patient Organisations</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Membership fees</td>
<td>7 000</td>
<td>7 862</td>
<td>-862</td>
</tr>
<tr>
<td>Eurordis In-Kind contribution</td>
<td>65 159</td>
<td>233 236</td>
<td>-168 077</td>
</tr>
<tr>
<td><strong>Volunteers</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Donations</td>
<td>987</td>
<td>150</td>
<td>837</td>
</tr>
<tr>
<td>In Kind Services</td>
<td>180 624</td>
<td>315 753</td>
<td>-135 129</td>
</tr>
<tr>
<td><strong>Health Corporates</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pharma. and Biotech Companies</td>
<td>574 108</td>
<td>616 648</td>
<td>-42 540</td>
</tr>
<tr>
<td><strong>Outside Health Corporates &amp; NPOs</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Outside Health Sector Corporates</td>
<td>75 000</td>
<td>55 965</td>
<td>19 035</td>
</tr>
<tr>
<td>Outside Health Sector NPOs</td>
<td>288 697</td>
<td>297 807</td>
<td>-9 110</td>
</tr>
<tr>
<td><strong>Miscellaneous</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Reimbursement</td>
<td>9 537</td>
<td>-</td>
<td>9 537</td>
</tr>
<tr>
<td>Others</td>
<td>3 896</td>
<td>3 071</td>
<td>825</td>
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<tr>
<td><strong>TOTAL INCOME</strong></td>
<td>1 205 009</td>
<td>1 530 490</td>
<td>-325 481</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>EXPENSES</th>
<th>2023 EUR</th>
<th>2022 EUR</th>
<th>Δ</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Staff</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Wages and charges</td>
<td>289 077</td>
<td>275 185</td>
<td>13 892</td>
</tr>
<tr>
<td>Other salaries</td>
<td>123 874</td>
<td>261 992</td>
<td>-138 118</td>
</tr>
<tr>
<td>Training and other costs</td>
<td>2 477</td>
<td>3 945</td>
<td>-1 468</td>
</tr>
<tr>
<td>Eurordis In-Kind Contribution</td>
<td>65 159</td>
<td>233 236</td>
<td>-168 077</td>
</tr>
<tr>
<td><strong>Logistics</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Travels and subsistence</td>
<td>126 549</td>
<td>118 984</td>
<td>7 565</td>
</tr>
<tr>
<td>Event logistics and catering</td>
<td>12 033</td>
<td>28 315</td>
<td>-16 282</td>
</tr>
<tr>
<td>In Kind services</td>
<td>180 624</td>
<td>438 821</td>
<td>-258 197</td>
</tr>
<tr>
<td><strong>Services</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fees</td>
<td>99 095</td>
<td>96 950</td>
<td>2 145</td>
</tr>
<tr>
<td>Reg Conf Partners</td>
<td>14 000</td>
<td>16 000</td>
<td>-2 000</td>
</tr>
<tr>
<td>Telecom and post</td>
<td>3 244</td>
<td>2 688</td>
<td>556</td>
</tr>
<tr>
<td>Other services</td>
<td>10 684</td>
<td>2 169</td>
<td>8 515</td>
</tr>
<tr>
<td><strong>Purchase</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Office furniture</td>
<td>2 635</td>
<td>1 940</td>
<td>695</td>
</tr>
<tr>
<td>Other purchases</td>
<td>1 072</td>
<td>2 724</td>
<td>-1 652</td>
</tr>
<tr>
<td><strong>Miscellaneous</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Financial expenses, Insurance, Tax</td>
<td>5 023</td>
<td>4 586</td>
<td>437</td>
</tr>
<tr>
<td><strong>TOTAL EXPENSES</strong></td>
<td>935 546</td>
<td>1 487 535</td>
<td>-551 989</td>
</tr>
</tbody>
</table>

**RESULT**                                    | 269 463  | 42 955   | 226 508 |
INCOME DIVERSIFICATION

- Patient Organizations: 1%
- Health Corporates: 48%
- In-Kind Contributions: 21%
- Non-Health Corporates & Non-Profit Organizations: 30%
Thank you to our members for contributing their images, stories, voices and quotes to the Annual Report 2023.

Special thanks to the following organizations for contributing pictures included in the report:

- Page 3 - Our Mission - Malaysian Rare - Malaysian Rare Disorders Society
- Page 2 - Our Impact - Instituto Unidos Pela Vida (Brazil)
- Page 16 - Building and Growing a Diverse Global Alliance - Ehlers Danlos Society, Illness Challenge Foundation (China), Rare Diseases Lesotho Association, Federación Mexicana de Enfermedades Raras (Mexico), World Alliance of Pituitary Organizations
- Page 21 - Strategic Engagement - Illness Challenge Foundation (China)
- Page 26 - High Level Forum - X Iberoamerican Meeting on Rare Diseases - Allianza Iberoamericana de Enfermedades Raras (ALIBER)
- Page 27 - APARDO Conference 2023 - Asia Pacific Alliance of Rare Disease Organisations (APARDO)
- Also a mention to EURORDIS - Rare Diseases Europe, Rare Disorders Kenya and the Wilhem Foundation for proposing images to be used in RDI communication

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