MEMBERSHIP MEETING 2022
16 JUNE, ONLINE
STRATEGIC ENGAGEMENT
Consultation and Work in Progress
RDI members express need and interest in building advocacy capacities

Member Feedback Survey

95% of participants would like assistance in helping build their capacity

Members highlight advocacy, global collaborations and access

<table>
<thead>
<tr>
<th>Topic</th>
<th>Interest</th>
</tr>
</thead>
<tbody>
<tr>
<td>National advocacy and rare disease national plans</td>
<td>60%</td>
</tr>
<tr>
<td>International advocacy and international collaborations</td>
<td>45%</td>
</tr>
<tr>
<td>Organizational development and sustainability</td>
<td>30%</td>
</tr>
<tr>
<td>Access to medicines</td>
<td>30%</td>
</tr>
<tr>
<td>Working with the WHO and international agencies</td>
<td>25%</td>
</tr>
<tr>
<td>Medicines, research, development and regulation</td>
<td>5%</td>
</tr>
<tr>
<td>Communication and social media development</td>
<td>5%</td>
</tr>
<tr>
<td>Digital health and assisted technologies</td>
<td>0%</td>
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</tbody>
</table>
NEW STRATEGIC ENGAGEMENT PROGRAMME
More comprehensive approach focused on advocacy and opportunities to engage and connect

Programme goals

1. Enable BROADER COMMUNITY to shape international collaborations
2. Support PATIENT ORGANISATIONS to drive local, national, and regional advocacy
3. Support COMMUNITY LEADERS
THREE KEY PATHS
Based on Members’ interests and RDI activities and priorities

- Advocating for PLWRD and their Families
- Participating in Global Collaborations
- Facilitating Access and Equity
PROGRAMME ELEMENTS

Resource Bank
Collection and organisation of existing community resources under the three key paths of interest

Toolkits and specific materials
New material to engage in RDI advocacy priorities
(#Resolution4Rare #UHC4Rare)

Events Calendar, Fellowships, Conferences
Calendar of community events, fellowships to participate and RDI Regional Conference Support

Member-led workshops and small group meetings
Opportunity to generate common knowledge on specific topics
(in 2021- CORD Advanced Therapies Series
In 2022 – RVA-RDI National plan workshop)

Tools and Materials
Engagement opportunities
CONSULTATION – “LET’S TAKE A TOUR”

► Content (what else would you like to see?)

► Layout (does the format make the information easy to understand?)

► Functionality (how easy is it to find information or resources?)
CONSULTATION – “TREASURE HUNT”

► Rare Disease Definition - How are rare diseases defined in the Philippines?

► When was India’s Rare Disease National Plan Adopted?

► Can you find the UHC4RareToolkit?

► Name 2 ongoing RDI Led Working Groups.
POLL – “RATE THE PLATFORM”

► **Content** *(Is the info useful? what else would you like to see?)*

► **Layout** *(Does the format make the information easy to understand?)*

► **Functionality /User Experience** *(how easy is it to find information or resources?)*
HOW CAN YOU HELP?

**Resource Bank**
Suggest resources on advocacy, global collaborations and access.

**Events Calendar, Fellowships, Conferences**
- Let us know about your upcoming conferences and events
- Include RDI as a media partners to your events
- Apply for fellowships

**Member-led workshops and small group meetings**
- Host a small group discussion
- Or request topic for a small group discussion
NEXT STEPS

2022

June
Member consultation
Feedback on digital platform. Call for resources and Member-led discussions

July-Sept
Crowd sourcing
Improve platform functionality & accessibility.

Sept - Oct
Programme Launch
Translate pages. Publish website. Host webinar on new programme

Oct - Dec
Develop & refine
Launch improved toolkits – Resolution & UHC

2023 and beyond
Grow
Continually refine programme. Strategic partnerships with other orgs to further develop programme

Aim is 2-3 Member-led Workshops in 2022
THANK YOU!
AGENDA

14:00 – 14:10
Opening Remarks and Message
Durhane Wong-Rieger, Council Chair, RDI

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Strategic Engagement of RDI Members
Consultation on New RDI Strategic Engagement Programme
Flaminia Macchia, Executive Director, RDI
Lisa Sarfati, Vice President, Community Engagement NORD
Hlawulani Mkhabela, Strategic Engagement Manager, RDI

15:00 – 15:10 (10 minutes break)

15:10 - 15:55
Advocacy Priorities
UN Resolution – Impact & Implementation
Next steps - UHC & Health Equity, the WHO and beyond
Flaminia Macchia, Executive Director, RDI
Dolores Cvitičanin, Public Affairs Manager, RDI
Roselyn Odero & Christine Mutena, Rare Disorder Kenya / Agnes Hactor, International Prader-Willi Syndrome Organisation / Anna MeriLuoto, Fabry International Network / Salome Mekhuzla, World Federation of Hemophilia

15:55 – 16:05 (10 minutes break)

16:05 -1 6:50
Global Access
Operational Description of Rare Diseases
IRDRC/RDI Access Task Force
Essential Medicines Lists & Essential Diagnostics List
Mary Wang, Science Policy Manager
Helen Malherbe, Rare Diseases South Africa / Rachel Yang, China Alliance for Rare Diseases / WHO Classifications, Terminologies and Standards Unit / Nuala Ryan, NCIRS Worldwide

16.50 - 17.00
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Durhane Wong-Rieger, Council Chair, RDI
MEMBERSHIP MEETING 2022
16 JUNE, ONLINE
ADVOCACY PRIORITIES
The Resolution & Beyond
TIMELINE

**Policy Event at the UN**
- 1st High-Level Event of the NGO Committee for Rare Diseases
- UN Headquarters, New York

**DECEMBER 2020**

**JANUARY 2021**

**Civil Society Key Asks & Draft Resolution**
- Civil Society proposal - RDI Advocacy Committee, RDI Council, RDI Members, EURORDIS & NGO Committee for RD

**7 JULY**

**Side event to the High-Level Political Forum**
- +300 Participants, 61 Countries
- Hosted by UN Core Group

**OCT & NOV:**
Informal negotiations

**15 NOVEMBER**
Adoption by Consensus of the UN Resolution by the 3rd Committee of UNGA

**1 MARCH**

**RDD Policy Event & 3rd High Level Event of the NGO Committee for Rare Diseases**
- Official launch of campaign for UN Resolution on PLWRD

**7-9 SEPTEMBER**

**UN Regional Meetings**
- 7 Regional meetings (6 UN Regions and EU)
- Attended by UN Reps

**16 DECEMBER**

Adoption of UNGA Resolution on Addressing the challenges of PLWRD
- Adopted by consensus
- 54 Co-sponsors
IMPACT OF THE RESOLUTION

- **Opportunity** - put rare diseases on the agenda of national policy makers and UN Permanent Missions
- **Strengthen the global RD movement** - shared mission and shared success as a community

Visibility - spotlight PLWRD who are invisible to society, policymakers and health systems

Recognition - recognise the challenges specific to living with a RD; and the need for particular attention & policies

Social Justice & Equity - RD go beyond health become a global equity priority

WHO - engage WHO & work toward a WHA Resolution - UHC for RDs

Other UN Agencies & Programmes within the UN System - work on more SDGs to address the holistic needs of PLWRD. This will advance the UN commitment to "leave no one behind"

National level - Global legitimacy & global framework to empower RD groups to drive advocacy at national level and pursue their specific priorities
INTERNATIONAL ADVOCACY PRIORITIES

1. UNIVERSAL HEALTH COVERAGE
   • Implementation of the Political Declaration on UHC

2. UN RESOLUTION
   • Prepare to “consider UN Resolution under Social Development”
   • Disability / Rare Diseases

3. WHO RESOLUTION
   • Preparing for a WHO Resolution in 2024

4. UN SYSTEM
   • Other UN Bodies linked with Key Asks of the UN Resolution and SDGs of Agenda 2030
INTERNATIONAL ADVOCACY TIMELINE (2021-2024)

16 December 2021
UNGA Resolution on Addressing the challenges of PLWRD

28 Feb 2022
 RDD Global Event World EXPO Dubai

24 May 2022
75th WHA Informal Side Event on Strengthening Health Systems for RDs

July 2022
Side Event at the HLPF on Gender Equality & Rare Diseases

28 Feb 2023
RDD Global Event

May 2023
76th WHA Informal Side Event on UHC for RDs

Sept. 2023
High-Level Meeting on UHC during UN High Level Week

Oct – Nov 2023
Consideration of Resolution under “Social Development” at the 78th Session of UNGA

May 2024
77th WHA Target date for a WHO Resolution on UHC for Rare Diseases

2021

2022

Work on the implementation of the UN Political Declaration on UHC

2023

Work towards the adoption of a WHO Resolution

2024

Follow up on the UN Resolution with UN Agencies and implementation at national level
RARE DISEASES AND DISABILITY

February
- Desk Research
  - To map existing resources on disability and rare diseases

March-May
- 27 Expert Interviews
  - To identify elements of differentiation and intersection

June-July
- Short Member Survey (open until 01 July)
  - To let members prioritize identified elements

July
- Webinar (19 July at 2 PM CET)
  - To present findings and conclusions to members and/or others to collect final feedback

July-August
- Research Report and Advocacy Strategy
  - To derive advice from research results
TOWARDS A WHO RESOLUTION

ONGOING:

• Benchmarking analysis of previous WHO resolutions

• **Mapping of countries** supportive of UHC and/or UHC4RD

• Draft document on the **process** towards a WHO resolution

• Note on the **instruments within the WHO** decision-making bodies and their respective legal capacities

- Maintaining constant relations with the **Core Group** (Brazil, Spain, and Qatar) and build relations with additional **co-sponsors** of the UN Resolution
- Building **multi-stakeholder support**, including with industry
RELEVANT UN BODIES

**General Assembly**
- **UNDP**
  United Nations Development Programme
- **UN-Women**
  United Nations Entity for Gender Equality and the Empowerment of Women
- **UNICEF**
  United Nations Children’s Fund

**SDGs**
- **SDG1** No poverty
- **SDG4** Quality education
- **SDG5** Gender Equality

**Secretariat**
- **OHCHR**
  Office of the High Commissioner for Human Rights

**Protection of human rights and full participation in society**

**Specialized Agencies**
- **World Bank**
  Global Fund for Rare Diseases?
- **ILO**
  International Labour Organization

**SDG8** Decent work and economic growth
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MEMBERSHIP MEETING 2022
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GLOBAL ACCESS
RD Description
Access to Diagnosis and Treatment
No universally recognized definition of rare diseases

... and no comprehensive description of rare diseases

Data adapted from Chan et al., 2020 and additional sources
Operational Definition of Rare Diseases

- RDI and WHO are collaborating under a Memorandum of Understanding “to improve the recognition of rare diseases and their visibility in health systems”, via development of an operational description of rare diseases.

- Operational definition is to be a policy document aimed at informing the WHO, and decision-makers when taking public health policy decisions.

- This document will give the frame for implementation of the Political Declaration on Universal Health Coverage and UN Resolution on addressing the challenges of PLWRD.
Expert Group

Ms Roberta Anido de Pena
Iberoamerican Alliance for Rare Diseases, Federación Argentina de Enfermedades Poco Frecuentes, Argentina

Dr Diego Ardigò
Chiesi Pharmaceuticals, Italy

Prof Gareth Baynam
Rare Care-Clinical Centre of Expertise for Rare and Undiagnosed Diseases, Perth Children’s Hospital, Western Australia

Prof Hugh Dawkins
DA Precision Health, Australia

Prof Ada Hamosh
Johns Hopkins School of Medicine, Online Mendelian Inheritance in Man (OMIM), USA

Dr Robert Jakob
Classifications and Terminologies Unit, WHO, Switzerland

Dr Eva Krpelanova
Classifications and Terminologies Unit, WHO, Switzerland

Mr Yann Le Cam
Rare Diseases Europe (EURORDIS), France

Dr Helen Malherbe
Rare Diseases South Africa, South Africa

Ms Jane Millar
International Health Terminology Standards Development Organisation (IHTSDO), SNOMED International, UK

Ms Caron Molster
Office of Population Health Genomics, West Australia Department of Health, Australia

Dr Lucia Monaco
International Rare Diseases Research Consortium, Italy

Prof Carmencita Padilla
University of the Philippines Manila, Philippines

Dr Anne Pariser
National Center for Advancing Translational Sciences (NCATS), NIH, USA

Dr Ana Rath
Orphanet, INSERM, France

Prof Peter Robinson
The Jackson Laboratory, USA

Prof Franz Schaefer
ERKnet European Reference Network, Heidelberg University Hospital, Germany

Dr Stefanie Weber
Code Systems, Federal Institute for Drugs and Medical Devices (BfArM), Germany

Ms Flaminia Macchia
Rare Diseases International

Dr Amy Whiting
Rare Diseases International

Ms Dolores Cvitičanin
Rare Diseases International
Structured consultation methodology

ROUND 0
Idea Generation
Identify elements for definition of rare diseases
Workshop 1
Nov 2021

ROUND 1
Assessment
Rate importance and acceptability of the elements
Survey 1

ROUND 2
Feedback & Discussion
Review and compare responses
Workshop 2
Dec 2021

ROUND 3
Reassessment
Review descriptors, structure of description framework
Survey 2

ROUND 4
Common Understanding
Review draft description of rare diseases
Workshop 3
Jan 2022
Operational Definition of Rare Diseases

Framework I. Challenges arising from rarity
Framework II. Recognising burden of rare diseases
Framework III. Catalyst for actions to address unmet needs

Core Definition
What is Rare
What is Disease

PART 1: CORE DEFINITION
PART 2: DESCRIPTIVE FRAMEWORK
CORE DEFINITION OF RARE DISEASES

PLWRD face distinct and significant challenges that arise from the infrequency of their medical conditions, such as a long diagnostic journey, inadequate clinical management, and limited access to effective treatments. The burden of rare diseases on patients, their carers and families, healthcare systems, and society overall, merits greater visibility and recognition.

A rare disease is a medical condition with a specific pattern of clinical signs, symptoms, and findings that affects fewer than or equal to 1 in 2000 persons living in any WHO-defined region* of the world.

Rare diseases include, but are not limited to, rare genetic diseases, rare cancers, rare infectious diseases, rare poisonings, rare immune-related diseases, rare idiopathic diseases, and rare undetermined conditions. While the frequency of most rare diseases can be described by prevalence, some rare diseases, such as rare cancers and rare infectious diseases, can be more precisely described by incidence.

**RARE**
1. Recognize PLWRD
2. Rarity defined by an epidemiological threshold
3. Recognize variables in geographical and country contexts
4. Towards a global recognition
5. Epidemiological thresholds for subtypes

**DISEASE**
1. Based on a distinct clinical presentation; alignment with International Classification of Disease
2. Irrespective of etiology
3. Irrespective of clinical severity
4. Recognize subclasses of rare diseases
5. Inclusive of conditions that remain undetermined

*WHO-defined regions are: Africa, Americas, Eastern Mediterranean, Europe, South-east Asia, Western Pacific*
Global Collaborations

Carine Alsokhn  Dr Eva Krpeleanova  Dr Helen Malherbe
Technical Officer  Medical Officer  Director of Research,
Classifications and Terminologies Unit, WHO  Rare Diseases South Africa
ICD-11

International Classification of Diseases
Eleventh Revision

ICD is a legally mandated global health data standard

ICD-11 is in effect since January 2022 (mortality and morbidity)

Independent of language and culture

Comparable statistics - semantic interoperability - for 150 years

ICD-11 is the most recent revision

- Full Integration of terminology and classification
- Fully digital, but paper use possible
- Scientifically updated
- All clinical detail codable, as much as desired

End-to-end digital solution (API, tools, online and offline)

Freely available through open license:
Creative Commons Attribution-NoDerivs 3.0 IGO (CC BY-ND 3.0 IGO)

All Rare diseases codable (URI or code)
How will the Operational Description of Rare Diseases support your work?

How can we support you?
GLOBAL ACCESS

Access to Diagnosis and Treatment
Universal Health Coverage & Essential Services

Target 3.8 | Achieve universal health coverage, including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all.
The new, phase 2 of the Working Group is coordinated by Rare Diseases International:

New IRDiRC-RDI Global Access Working Group was launched in October 2021.
Which are the essential medicines to treat RDs?

IRDiRC Essential list of medicinal products for rare diseases

- Published in July 2021
- 204 medicines extracted and organized in seven disease categories
- Living document to be regularly updated

The goal of the collated List is to further stimulate interactions between patient organizations, healthcare providers, industry and government agencies to improve standards of care for rare diseases and promote access to treatments.

IRDiRC – RDI Global Access Working Group Members

Durhane Wong-Rieger
Chair, RDI Council; President, APARDO; President & CEO CORD

William A. Gahl
National Human Genome Research Institute, NIH, USA

Alba Ancochea
Gareth Baynam
Marc Dooms
Kelly du Plessis
Michelle Erwee
Safiyya Gassman
Virgine Hivert
Ramaiah Muthyala
Daniel O’Connor
Manuel Posada
Domenica Taruscio
Samuel Wiafe
Scott Williams
Rachel Yang
Galliano Zanello
Flaminia Macchia
Ayda Ramazzina
Mary Wang

Alianza Iberoamericana de Enfermedades Raras (ALIBER)
Perth Children’s Hospital, Western Australia, Australia
University Hospitals Leuven, Belgium
Rare Diseases South Africa, South Africa
Takeda
Pfizer

EURORDIS, France
Indian Organization for Rare Diseases, India
Medicines and Healthcare products Regulatory Agency, UK
Instituto de Salud Carlos III, Spain
Instituto Superiore Sanità, Italy
Rare Disease Ghana Initiative, Ghana
Sanofi Genzyme
China Alliance For Rare Diseases, China
IRDiRC

Rare Diseases International
Rare Diseases International
Rare Diseases International
Identification of barriers to access

The goal is to identify common themes and challenges, and approaches on how the problems were solved / overcome in different countries.

- A bottom-up approach to gather real experiences and cases studies regarding access to medicines,
- Assess the roadblocks to access for different categories of medicines (e.g. chemical, biologics, generics, off-label, innovative, etc)
- Develop recommendations on how to overcome the roadblocks

Do you know whether X medicines are available in your country or region? Available via your health system, or specific insurance types?
Framework for Drug Journey

- Local Country Approval
- Logistics, Storage, Security
- Value Assessment, Price Negotiations
- Pilots, Usage Agreements, Contracts
- Access to Diagnosis, Specialist
- Availability to HCP
- Long-term Assessment, Evaluation, Usage & Access Update

- FDA / EMA Approval
- Supply Availability
- Special, Compassionate, Extended Access
- Awareness, Education
- Centre Treatment Capacity
- Consent, Monitoring, RWD Share
- Outcomes Assessment
Workstreams on Access

**ESSENTIAL MODEL LISTS**
- Updating of the IRDiRC List
- Referencing and promotion of the IRDiRC List
- Explore inclusion of RD medicines in the WHO’s Essential Medicines List
- Analysis of the Essential List of In Vitro Diagnostics

**DRUG ACCESS FRAMEWORK**
- Mapping of barriers in drug journey from manufacturing to delivery
- Identify potential roadblocks for different category of drugs
- Develop recommendations on how to overcome the roadblocks
Global Collaborations

Rachel Yang
Head of International Affairs,
China Alliance for Rare Diseases

Nuala Ryan
Chair of Trustees,
NCBRS Foundation
1. Literature Review and survey and systematic on existing models of telehealth,
   – uptake and usage by the rare disease community,
   – specific value and effectiveness,
   – identify the factors that enhance or limit their adoptability, sustained use,
   – efficiency/ease of access, and effectiveness in the rare disease community.
   – Models should include those used for provider to patient, and provider to provider communications, as well as for conducting research.

2. Identify barriers to and opportunities for the use of telehealth to improve access to diagnosis, care, and research experiences for rare disease patients
   – including technological, legal, cultural, linguistic, healthcare system, and patient/provider factors.

3. Develop “best practices” for introducing telehealth services into communities where they would be most beneficial using realistic and culturally-sensitive approaches, in partnership with local providers.
Why its important to me / Rare Disease Community

• For rare disease patients.
  – increased access to expert care and information

• For primary care clinicians/ Health Care systems
  – increased knowledge of treatments for rare diseases through a reciprocal educational plan
  – Sharing of information with specialists about the unique needs and cultural practices of their populations within that geographic region.
  – development of modules to treat rare diseases based on the gained knowledge

• For Researchers
  – access to patient populations with their rare disease of interest
  – use of this knowledge to understand natural history and develop new diagnostic, treatment, and management approaches..

• Payers, policy makers, and regulators
  – develop policies for this service as a legitimate aspect of care.
Q & A
THANK YOU!
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