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Rare Diseases in Latin America: A Dialogue with Experts

World Rare Disease Day

28 February, 2022



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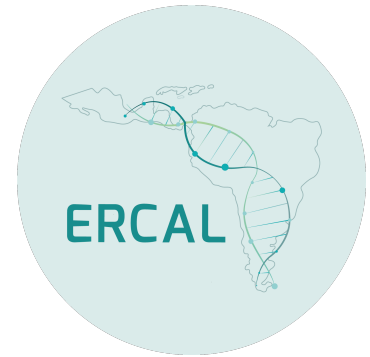


INDEX:

Introduction.....	3
Summary of the Event – Rare Diseases in Latin America: A Dialogue with Experts.....	5
Roundtable 1.....	5
<i>Panelists.....</i>	<i>6</i>
<i>Highlights.....</i>	<i>6</i>
<i>Conclusions.....</i>	<i>9</i>
Round Table 2.....	9
<i>Panelists.....</i>	<i>10</i>
<i>Highlights.....</i>	<i>10</i>
<i>Conclusions.....</i>	<i>12</i>
Publications.....	13



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Introduction

The event ***Rare Diseases in Latin America: A Dialogue with Experts*** was organized by “**Enfermedades Raras del Caribe y America Latina**” (ERCAL) in collaboration with the **Chan Zuckerberg Initiative** (CZI), and the **Americas Health Foundation** (AHF). Held on February 28, 2022- World Rare Disease Day- the event received the support from several patient association groups across the region, including Federación Argentina de Enfermedades Poco Frecuentes (**FADEPOF**), Federação Brasileira das Associações de Doenças Raras (**FEBRARARAS**), Federación Chilena de Enfermedades Raras (**FECHER**), Federación Colombiana de Enfermedades Raras (**FECOER**), Organización Mexicana de Enfermedades Raras (**OMER**).

Attendees:

Institutions:

- Heidi Bjornson representative of the Chan Zuckerberg Initiative (**CZI**)
- Stephen Groot, President of Rare Diseases of the Caribbean and Latin America (**ERCAL**)
- Richard Salvatierra President of the Americas Health Foundation (**AHF**)

Moderators:

- Felicitas Colombo Director of Public and Government Affairs of AHF
- Dr. Mariana Rico, Medical Director of AHF

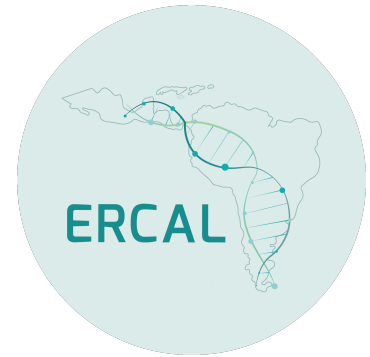
Patient Associations:

- Luciana Escati Peñaloza, Argentinean Federation of Rare Diseases (**FADEPOF**).





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- Antoine Daherm, Federação Brasileira das Associações de Doenças Raras (**FEBRARARAS**).
- Alejandro Andrade, Chilean Federation of Rare Diseases (**FECHER**).
- Diego Gil Cardoso, Colombian Federation of Rare Diseases (**FECOER**).
- Jesús Navarro, Mexican Organization for Rare Diseases (**OMER**).

Recordings of the event can be found in English, Spanish, and Portuguese via the below links:

English: <https://www.youtube.com/channel/UCUprLZYIWjgAB4VvznAg9QA/videos>

Spanish: https://www.youtube.com/channel/UCyH_1r5FyJG7b-BWMRH5-4w/videos

Portuguese: https://www.youtube.com/channel/UCMQm_x-cPJ2OAjU0kTa2cEQ/videos



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Summary of the Event – *Rare Diseases in Latin America: A Dialogue with Experts*

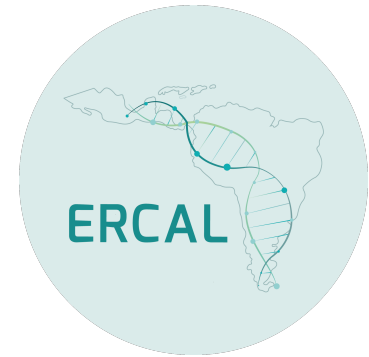
Roundtable 1

There are more than seven thousand rare diseases in the world and most of them have a genetic cause. According to Orpha.net approximately 4% to 8% of the world's population lives with a diagnosis of a rare disease, translating to 300 million people. This represents a significant group of people requiring specialized care in the provision of health services. In recent decades, there has been increased attention to efforts to stimulate research, development and access to medicines for rare diseases. Although there have been advances in public policy and regulatory frameworks, important reforms are still needed.

Currently, data on Rare Diseases in Latin America are scarce, the population of Latin America and the Caribbean according to the World Bank is 652,276,325 (650 million people). Based on this data, it is estimated that between 26 to 52 million people are living with a rare disease. Although progress has been seen, there are still regulatory, infrastructure, registration and public policy challenges, among others. The objective of the first roundtable was to 1) analyze the current landscape in Latin America, 2) assess the barriers and strengths in the region, and 3) propose recommendations to overcome these challenges.



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Panelists:

- Dr. Marisa Aizenberg (Argentina) – Academic Director of the Health Observatory at the U.B.A. Law School.
- Dr. Gustavo Mendes (Brazil) – General Director of Medicines and Biological Products at the National Health Surveillance Agency (ANVISA).
- Dr. Gabriela Repetto (Chile) – Director of the Rare Diseases Program at the Faculty of Medicine within Universidad del Desarrollo, Santiago de Chile.
- Dr. Germán Escobar Morales (Colombia) – Vice Minister of Health and Social Protection.
- Dr. Santiago March (Mexico) – Coordinator of Strategic Projects at FUNSALUD.

Highlights:

- The nomenclature of rare diseases varies across the Latin American region, including orphan diseases and diseases of low frequency.
- The need for a standardized definition for the term *rare diseases* across the region must be emphasized.
- A unified definition is important in terms of public policy, regulatory processes, access, research, and measuring intervention outcomes and reach.
- There must be an alliance with academia, which has a transcendent role in the transfer of technologies in the scientific and social spheres. In order to work together and seek solutions to the current barriers presented by rare diseases.



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- Although some countries have more advanced epidemiological data on RDs, it is still not possible to identify precisely which of the 8,000 RDs described by international organizations are found in each country.
- Patients' perspectives must be acknowledged, and their experiences must be addressed: What is their quality of life? What interventions can improve their quality of life? Which interventions are relevant in everyday life? Integrating patients, academia, industry, and regulators is key to creating collaborative research across traditional boundaries and improving patient journeys.
- It is important to emphasize the effective uptake of health services in order to increase the possibility of early disease identification. Although some countries have an epidemiological surveillance system, if primary health care professionals do not have diagnostic sensitivity, reducing the gap will be a much greater challenge.
- It is necessary to integrate different information systems – epidemiological surveillance and national registries of RDs that feed from such surveillance.
- Since it is difficult to guarantee the education of the entire medical community on RD, the adoption of scientific electronic records and the digitalization of health systems is being considered to generate systems that can alert health professionals when a RD should be suspected.
- Geographical, economic, social and cultural barriers, and a lack of specialists, result in a lack of access, prolonged processes to obtain health services, long waits for appointments.
- It is important to review and incorporate the possibilities offered by telehealth, telemedicine and teleassistance – collaborative learning platforms that are offered at the level of digital technology as a new way of thinking about health care. It is important to implement artificial intelligence for collaborative decision making, thus providing accurate and quality information to the patient.



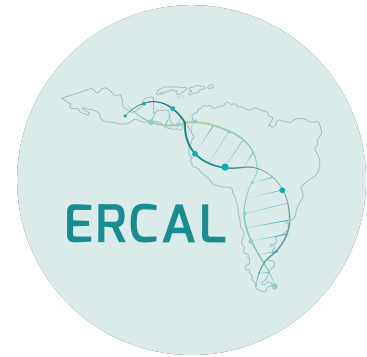
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- It is necessary to carry out health economics studies as well as cost-benefit studies for RDs to enable the eventual exploration of financing avenues.
- Generating region-specific data makes it possible to represent a reference point for the elaboration of public policies and the development of solutions. Evaluation models other than the traditional ones are needed to ensure that RDs are included among the important items in the benefits portfolio.
- It is important to not only encourage clinical and technology research, but also research on management models to facilitate care, reduce the fragmentation of the health system and the complexities of reaching rural and dispersed rural populations.
- Taking into account that around 70-80% RDs have a genetic underlying cause and that many of these are relatively easy to identify, it is important to generate information for the authorities to understand that opportune access to this technology results in personal, social and economic benefits for the health system. It is a myth that these processes are so expensive that they can defund health systems or people's finances. In fact, studies show that reducing the diagnostic odyssey by increasing the early use of genomic testing may be cost-effective.
- It is important to conduct a regulatory analysis of all countries. RD observatories where public policies are outlined describing their impact and mode of implementation should be developed. The shared experience of the processes through an observatory could be beneficial for the region as a whole.
- Below are some recommendations to improve the understanding and management of RDs in LA:
- 1. Increase training for first-level contact physicians and specialists
- 2. Create a National Molecular Diagnostic Platform
- 3. Implement National Registries



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- 4. Raise awareness on RDs

Conclusions:

As a region we have a great challenge. We have a long way to go in terms of diagnosis, registration, treatment, research and access in Latin America and the Caribbean. Although infrequent, altogether rare diseases generate a great impact on health systems as a whole and on the health of individuals. The solutions begin in spaces where cross-sectoral cooperation between regulators, governments, civil society, patient groups and the pharmaceutical industry is the focus, such as the one promoted by this roundtable. It is also of great importance to work in a coordinated way to face the challenges that surround RDs.

"We all deserve to live with dignity beyond our diseases, and equity is a fundamental pillar to living with more and better rights." - Dr. Marisa Aizenberg.

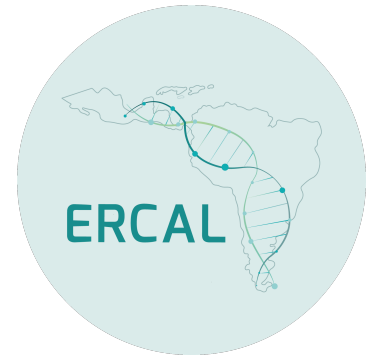
Round Table 2

Key points from the UN Resolution on Rare Diseases, published on December 16, 2021, in order to analyze the scope of its adoption in the Latin American region:

1. The inclusion and participation in society of people living with a Rare Disease and their families.
2. The guarantee of universal and equitable access to quality health services without financial hardship.
3. The promotion of national strategies and actions.
4. The integration of rare diseases into UN agencies, programs and priorities.



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5. Issuing periodic UN reports to monitor progress in the implementation of the resolution.

Panelists:

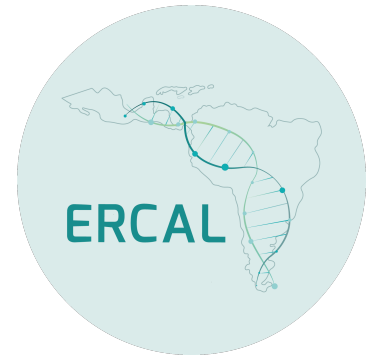
- Natalia Messina (Argentina) – Director of Specialty and High-Price Medicines at the National Ministry of Health.
- Melina Maia (Brazil) – Secretary of the Permanent Mission of Brazil to the United Nations.
- Dr. Manuel Espinoza (Chile) – Head of the Health Technology Assessment Unit of the Clinical Research Center of the Catholic University and Honorary Visiting Fellow of the Center for Health Economics at the University of York.
- Dr. Nubia Bautista (Colombia) – Deputy Director of Non-Communicable Diseases of the Ministry of Health and Social Protection.
- Dr. Tanya Palacios (Mexico) – legal director of Genes Latinoamerica AC and belonging to the Judicial Power of the Federation in Mexico.

Highlights:

The resolution established by the UN provides a groundbreaking recognition of the challenges that people living with RDs and their families face. It empowers the RD community at a global level and stimulates the development of national strategies and plans, but it is not enough. It is crucial that these regulations be implemented in order to reduce existing gaps and raise awareness to RDs but it is not enough. It is crucial that these regulations be implemented in order to reduce existing gaps and raise awareness to RDs.



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- The resolution is the basis for further integration of RDs into the UN priority action agenda that will serve as a guide for other countries to explore customized solutions to strengthen their policies to support RD needs. The resolution serves to legislation and increase society's awareness of RDs.
- This resolution recognizes the fundamental importance of equity, inclusion, and social protection mechanisms for people living with RDs, as well as calling for the elimination of discriminatory practices.
- The resolution recognizes the potential that children with RDs have. If children are provided with timely accurate diagnosis, adequate care and proper access to medicines, among other things, it would facilitate their integration into the activities of the State and participate in development.
- The resolution also recognizes the potential of civil society organizations to work with RDs. It identifies that there is potential for institutional change.
- The resolution addresses issues that go beyond health; it addresses employment, discrimination, vulnerability and gender issues. It also increases visibility to and understanding of those living with RDs.
- The resolution gives way to the unification of criteria at a regional level in order to address them in a more efficient and effective manner and with a more similar language.

How can progress be made in unifying the concepts of the regulations in the region and what are the challenges of implementing these regulations that have already been approved at the international and local levels?



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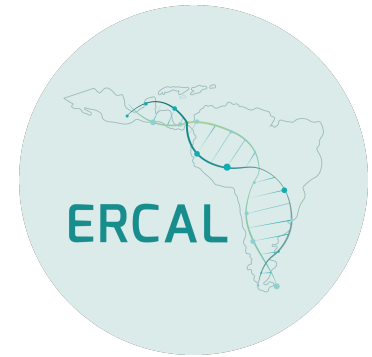
- The declaration provides a framework for thinking about the qualification and strengthening of specific legislation aimed at improving the right to health and other rights of people with RDs, their families and caregivers. Innovative elements are raised that automatically represent implementation challenges.
- Understanding and leading to implementation involves recognizing the particularities of the life course, specifically the impact on children and adolescents. It is also important to recognize the developmental environments: educational, community, and home. It turns out to be a challenge not only of enunciation but also of formulation and implementation of specific actions.
- It is important to link in a transversal and permanent way the psychosocial and mental health care of individuals and their families.

Conclusions:

Given the context and the new guidelines of the UN declaration, it would be timely to promote the integration of the region's initiatives and efforts through the coordination and leadership of a consolidated body. Throughout cooperation all these discussions can turn into action, it is important to take advantage of the visibility that the resolution has provided to RD. It is also relevant to share those guidelines that have already generated traction. This is further compounded by the traction generated by the pandemic that exposed the failures in health systems and the major impact these systems have on society and public health. The focus on health must be leveraged. The UN resolution provides a basis for increasing the visibility and rights of patients and how they should be incorporated into society. To take it a step further, it is necessary to innovate not only in technology, diagnostic methods and treatments, but also in implementing strategies and actionable mechanisms to give sustainability to care models and health systems. It should also be emphasized that the contribution and information that



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patient groups and civil society can provide for the creation of public policies and in the collection of data and research should also be enhanced.

Publications

Agencia EFE published a regional article based on an interview covering the event ***Rare Diseases in Latin America: Dialogue with Experts***

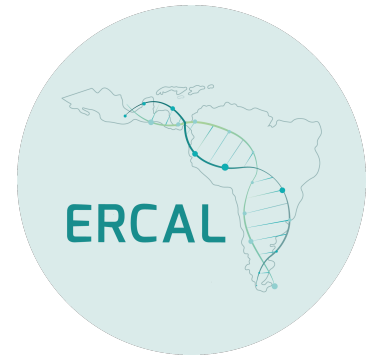
Latinoamérica enfrenta las enfermedades raras con múltiples carencias

EFE | Miami | 28 feb. 2022





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Link to the clip: https://www.efe.com/efe/america/mexico/latinoamerica-enfrenta-las-enfermedades-raras-con-multiples-carencias/50000545-4750226?utm_source=wwwefecom&utm_medium=rss&utm_campaign=rss