1ST CONGRESS ON Rare Diseases

LATIN AMERICA AND THE CARIBBEAN

March 16 & 17, 2023

Bogotá, Colombia

GENERAL REPORT

AHF AMERICAS HEALTH FOUNDATION

ERCAL ENFERMEDADES RARAS DEL CARIBE Y AMÉRICA LATINA

Chan Zuckerberg Initiative

IHS

Pontificia Universidad JAVERIANA Bogotá

FECOER Federación Colombiana de Enfermedades Raras

Partners

ADAPA

alsser
ALIANZA DE ASOCIACIONES DE ENFERMEDADES RARAS HUÉRFANAS Y POCO FRECUENTES DE PANAMÁ

FEBRARARAS

FECHER
FEDERACIÓN CHILENA DE ENFERMEDADES RARAS

FECOER
Federación Colombiana de Enfermedades Raras

feper
Federación Peruana de Enfermedades Raras

FERPOF
Federación Ecuatoriana De Enfermedades Raras O Poco Frecuentes

FIFARMA

omer
Organización Mexicana de Enfermedades Raras

Enfermedades RARAS CR

REMEXER
Red Mexicana de Enfermedades Raras

Prospectiva
PUBLIC AFFAIRS LAT.AM

Sponsors

GOLD

HORIZON

Biogen

Takeda

Chan Zuckerberg Initiative

AHF
AMERICAS HEALTH FOUNDATION

SILVER

illumina

ALEXION
AstraZeneca Rare Disease

RARAS CRO

sanofi

optimal therapies
A DIVISION OF salutis

ultragenyx

Boehringer Ingelheim

"There are more than 9000 Rare Diseases in the world (Orphanet), also known as orphan or complex diagnosis diseases. Most of them have a genetic cause. For size, it is estimated that approximately 3% to 8% of the world's population lives with a diagnosis of a RD, which means approximately 300 million people. This represents a significant group of people requiring specialized care in the provision of health services.

Currently, data on RD in Latin America is scarce. The population of Latin America and the Caribbean, according to the World Bank, is 650 million people. Based on this data, there are various estimates ranging from 20 to 52 million people living with a rare disease in Latin America."

Dick Salvatierra
CEO, Americas Health Foundation

Content

I. Introduction

Latin America and the Caribbean is ready to take action as a region, advance the rare disease (RD) landscape and improve the quality of life for those living with RD, their families and caregivers. This was the key message of the 1st Congress on RD for Latin America and the Caribbean, led by Americas Health Foundation and supported by partners and sponsors.

This historic event has set the stage for a new era of collaboration and cooperation in advancing the RE landscape in the region.

Through a patient-driven agenda, the Congress succeeded in increasing awareness, knowledge and participation of diverse stakeholders on key issues within the RD ecosystem, building the foundation for the next steps. Most importantly, it solidified the spirit, enthusiasm and desire of participants to advance collaborative efforts as a region.

This insightful event was led by AHF and supported by ERCAL, the Chan Zuckerberg Initiative, the Pontificia Universidad Javeriana, Federación Colombiana de ER (FECOER) and the Instituto de Genetica Humana; supported by 11 collaborating organizations (Adapa, Alasser, Febrararas, Fecher, Fecoer, Feper, Ferpof, Fifarma, Omer, Red ER and Remexer); and 11 sponsors (AHF, Biogen, Chan Zuckerberg Initiative, Horizon, Alexion, Boehringer Ingelheim, Illumina, Optimal Therapies, Raras Cro, Sanofi and Ultragenyx).

The congress featured 48 panelists from 12 countries, including leaders of patient organizations, policy makers, researchers, physicians, regulators, academics, government representatives, industry representatives and more. Some of the key opinion leaders who shared their perspectives at the event include Dr. Roberto Giugliani (Brazil), Dr. Gustavo Mendes Santos (Brazil), Dr. Ignacio Zarante (Colombia),

Dr. Enrique Teran (Ecuador), Dr. Natalia Messina (Argentina), Dr. Gabriela Repetto (Chile), Durhane Wong-Rieger (Canada) and Heidi Bjorson-Pennell (USA).
Throughout the 14 panels, experts delved into the reality of RD in Latin America and the Caribbean; the diagnostic odyssey, challenges in the patient journey, the importance of advancing genetic testing in RD, in having a holistic view of health workforce training for RD care in the region, differential approaches to health technology assessment in RD, innovative financing mechanisms to promote access and sustainability in RD care, the impact of digital health on RD, and more. Although resources, opportunities and needs vary across the region, Latin American countries face similar challenges to improve the management and comprehensive care of people living with RD. It was clear that Latin America must work together as a region, coordinate efforts, leverage lessons learned, promote education at all levels, and cooperate to continue to advance the RD landscape in the region.

Having mapped the existing opportunities, challenges and tasks ahead, AHF invites you to join us in carrying out key projects that emerged from this extraordinary event. Let's continue to work together to improve the quality of life for those living with RD, their families and caregivers in Latin America and the Caribbean. Together, we can make a real difference.

Dr. Mariana Rico
Medical Director, Americas Health Foundation

II. Panelists

Dr. Adriana Robayo 🇨🇴
Executive Director of the Institute for Health Technology Assessment (IETS), Colombia

Dr. Alejandra Vázquez 🇲🇽
Academic Director of the School of Medicine, Universidad Autónoma de Guadalajara, Mexico

Alejandro Andrade 🇨🇱
President of the Chilean Federation of RD - FECHER

Dr. Ana Cristina Ochoa 🇨🇴
Medical Director of Takeda.

Angela Chaves 🇨🇴
Patient Advocacy Senior Manager Region Intercontinental - Biogen

Antoine Daher 🇧🇷
Founder of Casa Hunter, Casa Dos Raros and President of the Brazilian Federation of RD, FEBRARARAS

Ariadne Dias 🇧🇷
Director of Institutional Relations of Casa Hunter - FEBRARARAS

Dr. Carlos Javier Alméciga 🇨🇴
Director of the Institute of Inborn Errors of Metabolism Pontificia Universidad Javeriana

Dr. Claudia Gonzaga J. 🇲🇽
International Laboratory for Human Genome Research, UNAM

Dr. Deborah Requesens 🇺🇸
Director of Jumpstart Program, Orphan Disease Center,
University of Pennsylvania

Dick Salvatierra 🇺🇸
Founder and CEO Americas Health Foundation

Diego Gil Cardozo 🇨🇴
President of the Colombian Federation of RD - FECOER

Durhane Wong-Rieger 🇨🇦
President and CEO of the Canadian RD Organization (Canada)

Dr. Elmira Safarova 🇨🇱
Co-Founder and CEO Rarus Health SPA

Dr. Enrique Terán 🇪🇨
Pharmacologist and President
of the Academy of Sciences of Ecuador

Felicitas Colombo 🇦🇷
Director of Government Affairs of AHF

Félix Galarza 🇪🇨
Executive President of the Ecuadorian Federation
of Rare or Infrequent Diseases (FERPOF)

Dr. Gabriela Repetto 🇨🇱
Director of the RD Program, Faculty of Medicine
of the Clínica Alemana Universidad del Desarrollo

Dr. German Escobar 🇨🇴
Former Vice Minister of Health and Social Protection of Colombia

Dr. Gustavo Mendes 🇧🇷
Former General Director of Medicines and Biological Products
at the National Agency of Health Surveillance, ANVISA

Dr. Heidi Bjornson-Pennell 🇺🇸
Chan Zuckerberg Initiative

Dr. Ignacio Zarante 🇨🇴
MD. President of the Colombian Association of Medical Geneticists -
Professor at the Institute of Human Genetics - Pontificia Universidad Javeriana

Irene Kanter Schlifke 🇳🇱
Director of Strategy and Customer Information
Biogen Digital Health

Lic. Jaqueline Tovar 🇲🇽
President of Mujer México and Founder
of Iniciativa pensemos en Cebras México.

Jesús Navarro Torres 🇲🇽
President of the Mexican Organization for Rare Diseases (OMER)

Dra. Juana Inés Navarrete 🇲🇽
President of the Mexican Association of Human Genetics

Dr. Leonardo Arregocés 🇨🇴
Former Director of Medicines and Health Technology
of the Ministry of Health and Social Protection, Colombia

Dr. Luis Pino 🇨🇴
Medical Oncologist, Founder and CEO of OxLER

Dr. Manuel Espinoza 🇨🇱
Head of the Health Technology Evaluation Unit
of the Clinical Research Center UC

María Elena Almendáriz 🇵🇪
Executive Director of the Peruvian Federation of RD, FEPER

Mariana Moreno Acevedo 🇨🇴
Spinal Muscular Atrophy Patient

Dra. Mariana Rico 🇨🇴
Medical Director of Americas Health Foundation

Mario Gómez 🇨🇴
Managing Partner, Prospectiva Consulting

Marisa Aizenberg 🇦🇷
Academic Director of the Health Observatory
of the Faculty of Law, University of Buenos Aires (UBA)

Dra. Marta Tamayo 🇨🇴
Director of the Right to Disadvantage Foundation and Professor
of the Institute of Human Genetics - Pontificia Universidad Javeriana

Dra. Natalia Messina 🇦🇷
Director of Specialty and High Price Medicines
of the Ministry of Health of Argentina

Dra. Olga Echeverri 🇨🇴
Associate Professor of IEIM - Pontificia Universidad Javeriana

Dra. Paula Daza 🇨🇨
Executive Director Center for Public Policy and Innovation
in Health (CIPS - UDD)

Dr. Reggie García Robles 🇨🇴
Medical Geneticist and Salubrista, Research Professor,
Pontifica Universidad Javeriana

Dr. Roberto Giugliani 🇲🇽
Co-President Casa Dos Raros

Roberto Rodríguez 🇩🇴
Vice President of the Dominican Alliance of Patient Associations

Ronny Garro 🇨🇷
Rare Disease Network Costa Rica

Dra. Tania Bachega 🇧🇷
President of the Brazilian Society of Neonatal Screening
and Congenital Errors of Metabolism

Vanessa Valencia 🇵🇦
Social Worker of aYOUdas Panama, ALASER

Yaneth Giha 🇨🇴
Executive Director, FIFARMA

III. **Historical Context**

of Rare Diseases (RD) in Colombia

María Belen Jaimes

Deputy Director of Non-Communicable Diseases, Ministry of Health and Social Protection of Colombia

In Colombia, RD are defined as those affecting 1 per 5,000 people. A brief account is given of the legal trajectory of these diseases in the country. Law 1392 of 2010, Law 1438 of 201, Law 1751 of 2015, National RD Roundtable - Resolution 1871 of 2021, Decree 1954 of 2012, Resolution 3881 of 2013, Decree 780 of 2016, SP Surveillance protocol, Resolution 946 of 2019, Resolution 651 of 2018, Decree 441 of 2022, Resolution 561 of 2019, Law 1960 of 2019 - Neonatal screening. Barriers that still exist in Colombia for RDs include: Diagnostic odyssey, implementation of neonatal screening, fractionated care, under-registration, lack of benefits for patients, and psychosocial and economic burden. The current government faces challenges related to RD in the country. Among these are the development and habilitation of referral centers, the creation of clinical practice guidelines (guidelines and protocols), improved access to genetic counseling, full implementation of neonatal screening, improvement of the information system and intersectoral management for the social protection of patients and their families.

See complete presentation HERE

IV. **Day 1**

March 16, 2023

1. Rare Diseases
The reality in Latin America and the Caribbean

PANELISTS

- **Dr. Ignacio Zarante**
  MD. President of the Colombian Association of Medical Geneticists - Professor at the Institute of Human Genetics - Pontificia Universidad Javeriana
- **Dr. Mariana Rico**
  Medical Director of AHF
- **Ariadne Dias**
  Director of Institutional Relations of Casa Hunter - FEBRARARAS
- **Alejandro Andrade**
  President of the Chilean Federation of RD - FECHER

MODERATOR

- **Felicitas Colombo**
  Director of Government Affairs of AHF

INTRODUCTION

This panel illustrates the situation of RD in the Latin American context. Each participant contributes from his or her place of experience, laying the groundwork for the conversation during the rest of the event.

The definition of RD varies globally and in Latin America there is no consensus on the definition of RD. Some countries such as Bolivia, Ecuador, Paraguay, Peru and Venezuela do not have a specific definition of RD. Others, such as Brazil, consider that RDs as defined by the World Health Organization affect 65 or fewer per 100,000 people. Argentina, Chile, Mexico, Panama and Uruguay adhere to the European Union definition of <1 per 2,000 persons affected and Colombia defines RD as affecting <1 per 5,000 persons. These diverse definitions also translate into the number of people living with a given RD varying across the region, depending on whether their condition meets different thresholds. The absence of a unified definition for the region creates challenges in estimating prevalence, creating standard policies and guidelines, integrating programs or registries, and allocating funds for research.

In addition, the maturity of the legislative landscapes of RD in each country varies, with some countries having specific laws for RD and others that do not yet have this type of legislation. However, our region also has very similar economic contexts, which determines the way in which health systems address this issue. Latin American countries face the common challenge of defining what a RD is, unifying the criteria for

defining these diseases, implementing solutions to guarantee early and accurate diagnosis, providing access to innovative diagnostic methods, considering available treatments, defining who will pay for these treatments and the task of generating public policies that consider the needs of caregivers. In terms of opportunities, there is the sharing of best practices in the approach to RD between countries (Casa dos Raros in Brazil), and analyzing the contributions of the study of RD for the benefit of common treatments.

KEY MESSAGES

1. RDs not only transform the lives of those who suffer from them, but also their family and social environment. From academia, governments and patient federations we have to think about protecting those patients with diseases that do not have a pharmaceutical laboratory behind them and need to be treated appropriately.
2. Regional coordination is necessary in order to homogenize RD policies, or at least bring them to a minimum understandable standard. The possibility of integrating RD into regional health systems presents the opportunity to update the entire health system with technology and knowledge that was not previously available.
3. It is important to think in a comprehensive manner about which RDs can really benefit from being on the relevant list of the ministries of health. For the countries of the region, the identification and recognition of RDs is very important, and it is just as important for an RD to be on the relevant list as for it not to be, since this creates barriers to access for other RDs.
4. Due to the similarity in socioeconomic issues of our Latin American countries, enabling channels of communication and exchange will always be of great benefit to the region.

Relevant information

- Presentation: Estimate of patients with RD in Latin America
- Presentation: Data on the notification of RD in Colombia
- Case Study: Casa Dos Raros
- Document: Consensus: Caregivers in Latin America

See the complete panel 1 HERE

2. Challenges in the patient's pathway
Testimonials of the diagnosis odyssey

PANELISTS

- **Ariadne Dias**
  Director of Institutional Relations of Casa Hunter-FEBRARARAS
- **Diego Gil**
  President of the Colombian Federation of RD - FECOER
- **Dr. Carlos Javier Almeciga**
  Director of the Institute of Inborn Errors of Metabolism - Pontificia Universidad Javeriana
- **Maria Elena Almendariz**
  Executive Director of the Peruvian Federation of RD, FEPER)
- **Vanessa Valencia**
  Social Worker of aYOUdas Panamá

MODERATOR

- **Felicitas Colombo**
  Director of Government Affairs of AHF

INTRODUCTION

This panel focused on the experiences of people with RD in Latin America. Presentations were given by leaders of patient organizations, who shared the challenges faced by patients and their families in accessing diagnosis, care and treatment, and the legal barriers they face. In addition, there were presentations by health professionals who discussed the care and access pathways for RD in Colombia as a case study.

The patient's pathway begins when the first symptoms are identified, which is why they should not be overlooked. It is not possible to have a comprehensive health system for RD when health services are fragmented, and even less so when they are designed for more prevalent diseases, which is why health systems must be strengthened starting with primary care, including all the actors involved. Regionalization of the evaluation systems is desirable, since there is a large amount of resources invested by each country in the generation of the same clinical practice guidelines and the same procedure manuals. In turn, sharing best practices and strengthening education on the processes of requesting new technologies can allow greater access to treatments and a reduction in the cases of judicialization of treatment requests, which only create greater barriers between regulatory bodies,

health systems and patients. From academia, RD centers and observatories can generate spaces where education, awareness and research on RD can be concentrated. The latter generates many benefits in itself, from the regionalization of information, to the strengthening of technologies and access to treatment for patients.

KEY MESSAGES

1. In many cases, the delay of a few days in the detection of a disease can be lethal for the patient; other diseases take between 3 and 15 years to be detected. The important thing is how timely that diagnostic confirmation is to ensure the quality of life of the patient with RD.
2. In the absence of a regional regulatory body, all countries in Latin America are often trying to regulate the same thing, independently investing a significant amount of resources to generate the same guidelines and procedures manuals.
3. There are a large number of diseases that are still undiagnosed, and the more technological tools we have, the greater the possibility of reaching an accurate diagnosis.
4. There are cases of laws in the countries of the region aimed at guaranteeing access to treatment for patients with RD. The challenges lie in the implementation of these laws, since the judicialization of the processes for requesting access to treatment does not benefit any of the stakeholders.
5. RD can define the model of differentiated care, especially considering that there are no formal guidelines on care pathways for patients with RD.
6. In addition to work on strengthening primary care in order to channel the patient from the clinical suspicion, it is very important to work on guaranteeing access to timely and adequate diagnosis in order to have access to adequate treatment in those cases where it exists, since the therapeutic offer for RD is very low, for which reason it is also convenient to promote clinical research.

Relevant information

- Orphan Disease Center , UPENN, USA
- Observatório de Doenças Raras, UBR, Brazil
- Institute of Inborn Errors of Metabolism, Universidad Javeriana, Colombia.

See complete panel 2 HERE


AHF
ERCAL
Chan Zuckerberg Initiative

3. Molecular and Genetic Diagnosis of RD: A call to action

PANELISTS

- **Dr. Gabriela Repetto**
  Director of the RD Program, Faculty of Medicine of the Clínica Alemana Universidad del Desarrollo
- **Dr. Claudia Gonzaga Jauregui**
  International Laboratory for Human Genome Research, UNAM
- **Dr. Enrique Teran**
  President of the Academy of Sciences of Ecuador
- **Dr. Carlos Almeciga**
  Director of the Institute of Inborn Errors of Metabolism, Pontificia Universidad Javeriana

MODERATOR

- **Dr. Mariana Rico**
  Medical Director AHF

INTRODUCTION

The panel focused on the latest advances in the molecular and genetic diagnosis of RD in Latin America. The panel featured experts in orphan diseases who shared their knowledge and experience on the diagnostic process, the genetic and molecular basis of these diseases and current and emerging technologies for their diagnosis. It also included discussions on the challenges and barriers to access to molecular and genetic diagnostics in Latin America and proposed strategies to increase access to these diagnostic tools for patients and families affected by RD.

GENERAL CONCLUSIONS

A predominant challenge surrounding RDs is the difficulty of reaching a timely and accurate diagnosis. RDs vary in their definition between countries, and this generates discrepancies in diagnosis and access to treatment. In addition, there is a lack of knowledge in the medical community about the early symptoms and diagnosis of RD, which also leads to delays in their detection. If we consider that 80% of RD have a genetic component and the genetic cause of more than 6000 diseases is known, we are missing an important opportunity in terms of diagnosis, especially when the technology to reach it already exists. And while genetic sequencing is an excellent avenue for diagnosis, the initial clinical practice cannot be overlooked, as it is the first

step in the patient pathway. Increasing access to genetic sequencing and sharing information regionally helps to reduce variants of uncertain significance to the benefit of not only RDs, but the healthcare system as a whole. However, there are major challenges that impede the implementation of molecular diagnostics in Latin America and the Caribbean, such as lack of knowledge and support for its implementation by government and decision-makers, limited coverage by public and private health systems, few specialized laboratories and lack of trained personnel, high costs and limited access, low demand due to high costs and low access, and lack of knowledge of the advantages of molecular diagnostics by physicians. The participation of all stakeholders is required for the implementation of effective diagnostic programs, and the possible alliance with other not so obvious stakeholders (such as Genome Canada) to facilitate the use of technology and installed capacity. A program focused on genetic counseling or accompaniment for patients and families, where the counselors are primary health care professionals, is also recommended.

KEY MESSAGES

1. The diagnostic odyssey refers to the path that the patient travels in his or her search for an accurate diagnosis. In Latin America, the average time for diagnostic confirmation of a RD is 10 years, while in countries such as the United States and some European countries, where molecular diagnosis is one of the first options for confirming suspected genetic diseases, the time has been reduced to an average of 4 to 5 years.
2. Since the 1970s, with the karyotype karyogram, a diagnostic certainty of approximately 3% has been achieved. With current technological advances, diagnostic certainty increases to approximately 50%. Although genetic sequencing has changed the diagnostic testing scenario, treating physicians continue to wonder which test is appropriate for the RD patient they are treating.
3. There are 3 fundamental elements that interact around diagnostic tests: on the one hand, infrastructure, on the other hand, human talent and finally, financial resources. Latin America has a lack of infrastructure since there are no policies for acquiring the necessary equipment and prices tend to be very high, there are few trained professionals and they are generally concentrated in large cities, and finally, no resources are allocated by decision-makers to ensure that diagnostic testing is a sustainable strategy.
4. The countries of the region take as a reference genomic data made with European population and not having Latin American data as a reference increases the possibility of obtaining variants of uncertain significance.

5. It is necessary to vindicate the role of a good physical examination and a good analysis of the family history to be able to approach an accurate diagnosis, in addition to accepting what is known and what is not known in order to be able to use the correct tools in the search for diagnosis.
6. The creation of a regional training program on Genetic Counseling and the survey of installed capacities for the generation of genetic information by country can strengthen and promote genetic diagnosis and significantly reduce the time in the diagnostic odyssey of patients with RD.

Relevant Information

- Publication Accelerating Access to Genomics For Global Health
- Web Site: Genome Canada
- Web Site: National Cancer Law (Chile)

See complete panel 3 HERE

4. Advancing Neonatal Screening in Latin America.

PANELISTS

- **Dr. Claudia Gonzaga Jauregui**
  International Laboratory for Human Genome Research, UNAM
- **Dr. Ignacio Zarante**
  MD. President of the Colombian Association of Geneticists, Professor Instituto de Genética Humana - Pontificia Universidad Javeriana
- **Dr. Tania Bachega**
  President of the Brazilian Society of Neonatal Screening and Congenital Errors of Metabolism
- **Dr. Juana Inés Navarrete**
  President of the Mexican Association of Human Genetics
- **Lic. Jacqueline Tovar**
  President of Mujer México and Founder of Iniciativa pensemos en Cebras México.

MODERATOR

- **Dr. Mariana Rico**
  Medical Director of AHF

INTRODUCTION

This panel focused on the current status and development of newborn screening programs in Latin America and their importance in the public health context. The panel featured experts in the field who shared their knowledge and experience on legislation and policies related to newborn screening, current and emerging technologies for screening, and the challenges and barriers to access to newborn screening in the region. The panel included discussions on the potential benefits of expanding newborn screening programs, including early identification and intervention for RD. The panel provided a valuable opportunity for attendees to learn about the latest advances in newborn screening and to discuss the challenges and opportunities for improving access and legislation related to newborn screening in Latin America.

GENERAL CONCLUSIONS

Newborn screening is a valuable public health strategy performed in newborns to identify potentially serious diseases before the onset of symptoms or early enough to warrant therapeutic intervention, reducing morbidity and mortality and improving

quality of life. Newborn screening is an essential vanguard for infant care and has provided great improvements in the early diagnosis of many congenital diseases. However, of the 33 countries in the region, only 16 have a neonatal screening program that screens for 5 to 6 diseases, some screen for fewer diseases, and some have no national neonatal screening programs. Within this varied landscape of newborn screening programs and legislation are also different degrees of implementation of the laws. For example, Colombia and Brazil passed new legislation to expand newborn screening programs, however, they face challenges in the effective implementation of such programs. Some of these challenges include geographic coverage, post-screening follow-up of patients, informed consent, and ethical issues surrounding screening (e.g., if treatment is not available or covered).

Therefore, in addition to the regulatory framework, consideration should be given to the necessary budget for a gradual growth of neonatal screening detection, since operability and uniformity in screening guarantee the success of the programs while contributing to disease statistics in the region. Neonatal screening should be treated as a public health priority, and the data uncovered can attract research to countries in the region, thereby increasing the capabilities of each country and facilitating the exchange of best practices. It is also important to promote education of the general public about the importance of neonatal screening and to ensure effective communication between the physician and the parents of the baby.

KEY MESSAGES

1. Latin American countries still have a long way to go in terms of neonatal screening, because although some countries have active legislation on screening, the fragmentation of health systems also generates differences in the number of diseases to be identified.
2. While there is an advantage in identifying diseases in private screening programs, most of the time, when a pathology is identified, patients turn to the public system. This is one of the reasons why decision makers should invest in newborn screening.
3. In the face of the constant argument that there are no resources to invest in expanding screening panels, we must show that it is more cost-effective to detect than to treat the sequelae of late diagnosis.

Relevant information

- Figure: Comparison of Screening in Latin American Countries
- Video: Think Zebras Initiative

See complete panel 4 HERE


1ER

5. UN Resolution on RD in Latin American Context

PANELISTS

- **Durhane Wong-Rieger**
  President and CEO of the Canadian RD Organization (Canada)
- **Dr. Natalia Messina**
  Director of Specialty and High Price Medicines of the Ministry of Health of Argentina (Argentina)
- **Dr. German Escobar**
  Former Vice Minister of Health and Social Protection of Colombia (Colombia)
- **Dr. Gustavo Mendes**
  Former General Director of Medicines and Biological Products at the National Agency of Health Surveillance, ANVISA (Colombia)

MODERATORS

- **Felicitas Colombo**
  Director of Government Affairs AHF
- **Dr. Mariana Rico**
  Medical Director of AHF

INTRODUCTION

This panel focused on the United Nations Resolution on RD (UN RD Resolution) and its implications for the Latin American region. The panel brought together experts from the RD field, policy makers and UN representatives to discuss the UN Resolution on RD and its impact on the Latin American region. It examined the progress made in the implementation of the resolution and the challenges that still need to be addressed. Possible solutions to improve the diagnosis, treatment and care of people with RD in Latin America were also explored.

GENERAL CONCLUSIONS

On December 16, 2021, the UN adopted the first-ever resolution on addressing the challenges of people living with a RD and their families. Recognizing the need to promote and protect the human rights of all people, including the approximately 300 million people living with a RD worldwide. The main objective of the UN Resolution on RDs is to recognize RDs as a global public health issue, not only in the health field, but also in the socio-cultural, community, etc. environment. The implementation of the UN

Resolution on RD has been a challenge at the global level and especially in Latin America, since it establishes that countries will support initiatives that allow the recognition of RD, and this in Latin America represents a challenge in itself due to the lack of infrastructure, specialists and financial resources for this purpose.

When a health system establishes its own definition of what a RD is, as in the case of Argentina (1 in 2000 cases), it "imports" prevalence data from other countries, since there is no local registry. Not having a unified definition as a region is the greatest challenge for the identification of patients and the regional approach to this problem. According to Durhane Wong-Rieger, President and CEO of the Canadian Organization for RD, less than 1% of patients who are eligible for treatment are able to access them due to multiple barriers to treatment access. If we consider that most pharmaceutical companies base the cost of their treatments on the expectation of accessing 10% of eligible patients, then we must add industry to the access conversation, as other models can be enabled that consider access to treatments for a higher percentage of patients, such as the Payment by Results model implemented in Argentina on the first approved Gene Therapy.

In this way, treatment costs could be reduced for the patient or the health system under the premise of having a higher volume. The strengthening of public policies at the local level is necessary to establish a national registry that will generate data to guide and support the development of future public policies and where the efforts made to subscribe to the resolution can be evidenced. This is necessary, since the current resolution presents a challenge that no one anticipated, and that is to have

to renew the resolution, with the data provided by each of the member countries of the United Nations. There is also another resolution on the way, which is that of the World Health Organization. Unlike the UN resolution, those who adhere to the WHO resolution will have to ensure implementation and patient access to multiple services. As a region, countries have a lot in common, and sharing more than just best practices, but also big mistakes and areas of opportunity can help other countries avoid them.

KEY MESSAGES

1. RDs are a global public health issue, not only in terms of health, but also in social, economic, educational and employability terms.
2. While disability and childhood resolutions already exist, patients with RD are at an even greater disadvantage as they are often not covered by these programs.
3. The resolution does not solve the problem, but by creating the space for recognition of RDs, it allows progress to be made in access to diagnosis and treatment for patients with RDs.
4. The resolution not only addresses health issues, but also takes into account the integral nature of the patients and their families and allows for a differentiated approach, focused on the needs of each patient.
5. There is no linear route for the implementation of the resolution in the countries of the region; however, there are elements that every country needs to take into account for intersectoral implementation; first and foremost, data on RD, with information systems that are gradually evolving to integrate data beyond the health sector.
6. The biggest challenge regarding the UN resolution on RE is to go back and renew it, and for this, we must answer what was done with the established agreements and prove whether they effectively generated or initiated the expected changes.

Relevant information

- Web Page: UN Resolution on RE
- Web Page: ER Registration System in Argentina
- Web page: Introduction of the first gene therapy in the Argentine health system.

See complete panel 5 HERE

6. Case Study

Regional integration of centers of excellence for comprehensive and differential care of RD

PANELISTS

- Dr. Roberto Gugliani
  Co-President Casa dos Raros
- Dr. Ignacio Zarante
  President of the Colombian Association of Medical Geneticists, Professor at the Institute of Human Genetics - Pontificia Universidad Javeriana
- Antoine Daher
  Founder of Casa Hunter, Casa Dos Raros and President of the Brazilian Federation of RD, FEBRARAS

MODERATORS

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

INTRODUCTION

This panel focused on the integration of centers of excellence for RD in the region, highlighting the successes and challenges of these initiatives. The panel brought together RD experts to present a case study of successful integration of centers of excellence in Brazil and discussed the challenges faced in implementing these initiatives. The potential for replicating these efforts in other Latin American countries was also explored. The panel provided valuable information for those interested in RD and efforts to improve care for affected people in Latin America. In addition, the panel helped to promote the exchange of knowledge and best practices among different countries and regions in Latin America, encouraging the development of a comprehensive and differential approach to the care of these diseases.

GENERAL CONCLUSIONS

In the Latin American region, Brazil is the country that invests the most in health with an expenditure of 13% of its GDP in 2022. To address the initial challenges in RD, genetic diagnosis and follow-up was done in university hospitals, to cover these expenses from research and education budgets.

The Hospital de Clínicas de Porto Alegre developed the first reference center, and created the first training and information systems for RD in Brazil. In 2016, the government began designating referral services for RD. There are currently 21. These centers have services that are not very standardized among themselves and are still few in relation to the population and their geographic distribution, being mostly concentrated in major cities . The most recent initiative in Brazil is Casa Dos Raros, a reference center inaugurated on February 28 (World RD Day). It is a center specialized in comprehensive care and research for patients with RD. There is an initiative to replicate this center in Colombia, based on a resolution that contemplates 23 criteria to be met for a care center to be considered a reference center. Currently there are only 4 centers authorized to provide services for Multiple Sclerosis. At the Hospital Universitario San Ignacio, in Bogota, Colombia, work has begun to set up this center, taking into account the challenges involved.

Relevant information

- Presentation: RD in Brazil
- Web Page: RD Network Brazil
- Website: Medical Genetics Information Service
- Web Page: Casa dos Raros

See complete panel 6 HERE

7. Mariana Moreno Acevedo

Testimonial of patient living with a RD

PANELISTS

- **Mariana Moreno Acevedo**
  Spinal Muscular Atrophy Patient

MODERATORS

- **Felicitas Colombo**
  Director of Government Affairs AHF
- **Dr. Mariana Rico**
  Medical Director of AHF

DESCRIPTION

Mariana is a patient of Spinal Muscular Atrophy, and at 21 years old, she shares how not only the physical impairments generate the disability of patients, but also the entire environment. Although the first symptoms manifest themselves at the age of 1 year, it is not until the age of 16 that she was able to access comprehensive treatment, even after going through specialists who continued to misinterpret her condition and despite having a genetic test that confirmed. This is just one example of many in which 15 years becomes the standard time between the first symptoms and access to treatment.

Additional information

- Video testimonials from three patients in Mexico, Ecuador and the Dominican Republic: https://youtu.be/zNLiRCnlNMU?t=7352

See complete panel 7 HERE

8. Holistic View of education and training of health care personnel in RD

PANELISTS

- Dr. Alejandra Vazquez
  Academic Director of the School of Medicine, Universidad Autónoma de Guadalajara, Mexico
- Dr. Marta Tamayo
  Director of the Right to Disadvantage Foundation and Professor of the Institute of Human Genetics - Pontificia Universidad Javeriana
- Dr. Olga Echeverri
  Associate Professor of IEIM - Pontificia Universidad Javeriana
- Dr. Enrique Terán
  Pharmacologist and President of Sciences of the Academy of Ecuador.

MODERATOR

- Felicitas Colombo
  Director of Government Affairs AHF

INTRODUCTION

This panel focused on the importance of training health professionals in the diagnosis, treatment and care of people with RD. The panel brought together experts to discuss the current state of health personnel training in the region and the challenges that need to be addressed. Successful training programs were presented, highlighting best practices and the benefits that can be derived from such initiatives. Possible solutions to improve the diagnosis, treatment and care of people with RD in Latin America were also explored. In addition, the panel addressed the importance of a holistic approach in the training of health professionals, including not only technical knowledge but also social and emotional aspects.

GENERAL CONCLUSIONS

The main barrier to having a holistic vision in health education is precisely that there is no holistic vision in the academic training programs of universities in the region. The RD subject is not common in health training curriculum, and given the technological advances in health, new subjects have been included, prioritizing science over soft skills. Another challenge is when students face the reality of clinical practice, where they only have about 15 to 20 minutes to bond with each patient in a regular day. It is important to share and replicate the sensitization models that are currently

being carried out in the region, and to also sensitize general care personnel, not just health personnel. This means taking the subject out of the academy and teaching in the field, while insisting on an academic change where not only health decision-makers but also educational decision-makers are involved.

The importance of RD education in the training of general practitioners is that they are the ones who are likely to become the first point of contact for a patient presenting symptoms of an RD, whether in a primary care setting, pediatric practice, etc. Many patients and their families visit multiple physicians during the diagnostic odyssey, obtaining erroneous diagnoses and delaying the possibility of treatment and therapy. It is important to emphasize that not all physicians need to know about all RDs or have the ability to diagnose and treat them. However, they should be able to recognize which symptoms may raise the suspicion of an RD and be able to refer that patient to an appropriate specialist for diagnosis.

KEY MESSAGES

1. Medical training programs do not include the part where they explain and sensitize the student to the fact that the patient is, first and foremost, a person. Such preparation and sensitization can contribute to improving patient communication skills.
2. The academic environment is, above all, the place where opportunities are generated for the analysis of medical cases that may lead to the confirmation of a diagnosis of RD.
3. Implementing a holistic view through additional classes in the training of physicians is a difficult challenge to address, since the medical specialty in topics and knowledge has been expanding, but career time has not.
4. Communication in medicine consists of teaching the student and future physician about how to transmit information to their patients, how to generate the necessary empathy and how to recognize on a personal level the difficulties of inclusion that patients with RD go through.

Relevant information

- Web Page: Right to Disadvantage Foundation

See complete panel 8 HERE

9. La experiencia de las organizaciones de pacientes en Latinoamérica y el Caribe

PANELISTS

- Vanessa Valencia Barroso
  Social Worker of aYOUdas Panama, ALASER
- Diego Gil Cardozo
  President of the Colombian Federation of RD - FECOER
- Ariadne Dias
  Director of Institutional Relations of Casa Hunter - FEBRARARAS
- Roberto Rodriguez
  Vice President of the Dominican Alliance of Patient Associations
- José de Jesús Navarro Torres
  President of the Mexican Organization for Rare Diseases (OMER)
- Alejandro Andrade
  President of the Chilean Federation of RD - FECHER
- Félix Galarza
  Executive President of the Ecuadorian Federation of Rare or Infrequent Diseases (FERPOF)
- Maria Elena Almendáriz
  Executive Director of the Peruvian Federation of RD, FEPER
- Ronny Garro
  Rare Disease Network Costa Rica

MODERATOR

- Felicitas Colombo
  Director of Government Affairs AHF

DESCRIPTION

This panel focused on the experiences of orphan disease patient organizations in the region, highlighting the successes and challenges of these initiatives. The panel brought together leaders of RD patient organizations to share their experiences and insights. It provided valuable information for those interested in these diseases and efforts to improve care for people affected by these conditions in Latin America, featuring the voice and perspective of the patient and highlighting the importance of the patient-centered approach in the management of RD.

The following links show the incidence data, registration information, codification of RD, regulatory framework, patient organizations, representation of these

organizations, national research programs, specialized centers, initiatives, opportunities and challenges in each of the countries of the region.

KEY MESSAGES

1. The region does not have a unified definition of RD.
2. Not all countries have specific legislation on RD.
3. There is no single registry of RD in Latin American countries, so the data available in the region is very scarce.

Brasil
Chile
Colombia
Costa Rica
Ecuador
México
Panamá
Perú
Republica Dominicana

See complete panel 9 HERE

V. **Day 2**

March 17, 2023

10. Health technology assessment for RD with differential approach

PANELISTS

- Diego Gil Cardozo
  President of the Colombian Federation of RD - FECOER
- Dr. Paula Daza
  Executive Director Center for Public Policy and Innovation in Health (CIPS - UDD)
- Dr. Adriana Robayo
  Executive Director of the Institute for Health Technology Assessment (IETS) Colombia
- Dr. Leonardo Arregoces
  Former Director of Medicines and Health Technology of the Ministry of Health and Social Protection, Colombia
- Dr. Gustavo Mendes
  Scientific Researcher at IVI - Former General Manager of medicines and biological products at ANVISA.

MODERATOR

- Dr. Mariana Rico
  Medical Director of AHF

INTRODUCTION

This panel focused on patient participation in Health Technology Assessment (HTA) processes as a tool to evaluate the clinical, economic and social impact of new RD technologies in Latin America, taking into account the specific characteristics and needs of patients. This panel brings together experts in the field of HTA and RD, who share their experience and best practices to evaluate the effectiveness, safety and cost-effectiveness of new technologies, as well as their social implications and the ethical issues they may raise.

GENERAL CONCLUSIONS

Health technology assessment (HTA) for RD poses challenges to traditional methodologies. The nature of these diseases means that there are limitations in the number of patients in studies, the time to onset and diagnosis, and the approach to precision medicine. Therefore, there is a need for a broader evaluation framework, which should consider not only effectiveness and safety, but also equity, patient perspectives, and additional costs.

The lack of formalization of evaluation processes is one of the main barriers to the involvement of patient organizations. In addition, there is limited communication between evaluation agencies and the patient community, which hinders the exchange of information and best practices.

Brazil has a formal patient participation process for HTA, with three formal spaces for social participation: patient experience, consultation and public hearing. The process in Brazil is transparent, but there is a need to encourage patient participation while preserving and guaranteeing patient privacy. The Brazilian committee privileges the quality of the social contribution over quantity, which means that, even if there are 40,000 contributions for the incorporation of a drug, the decision will not necessarily be positive if the contributions are not of high quality.

The Ricarte Soto Law in Chile is a legal framework that enables patient participation in HTA for RD, providing a more rational and organized approach. This law has particularities that allow the inclusion of RD in the prioritization commission, where patient groups participate in the evaluation of the social impact of the technology.

The use of real-world evidence is a challenge for HTA for RDs, as some regulatory agencies do not accept this information. This poses a challenge to understanding the effectiveness of RD treatments and making informed decisions about market continuation, coverage, and other factors. Agencies that are willing to communicate and dialogue with industry and other stakeholders are more likely to achieve a differential HTA. Brazil is cited as a prime example of such an agency in Latin America. The cost of RD studies is high, so it becomes imperative to design effective and useful studies for future decision making. Patient perspectives and expectations must be taken into account in study designs and results to ensure that they are representative of real-world scenarios. To achieve this, collaborative discussions and agreements between regulatory agencies, technology evaluators and industry stakeholders are needed.

Latin America needs more people who understand and are trained in RD and HTA. While progress has been made in educating patients and involving them in the evaluation process, more professionals need to be involved in these discussions. Leadership is key to effective collaboration between agencies, patients, and industry when it comes to differential HTA for RDs. This leadership must come from the highest levels of government and regulatory agencies, and must focus on changing the way things are traditionally done. The leader must be someone who is willing to engage in dialogue and listen to patients and industry representatives without being condescending. This type of leadership should focus on changing the way agencies interact with different stakeholders, particularly patients, and should be based on a willingness to engage in dialogue and understand different perspectives. Change in leadership should not be seen as a loss of control or governance. Rather, it should be seen as a way to improve the regulatory process and ensure that patients receive the best possible care. Ultimately, the goal of differential HTA for RDs is to improve patient outcomes, and effective leadership is key to achieving this goal.

KEY MESSAGES

1. Patient and citizen participation in the HTA process for RD is fundamental for a differential approach. They understand the value of the technology, its benefits and outcomes. Communication and coordination channels between regulatory agencies and patient organizations are necessary to strengthen the generation of evidence and build these processes with objectivity. Patient organizations have great potential to generate evidence and contribute to these spaces, but it is crucial to understand the role, purpose and scope of each one in the different phases of HTA.

2. The quality and organization of patient participation is crucial to the success of HTA interventions. Patients and their representatives should be empowered to understand the evaluation process. There is an opportunity to formalize processes for active patient and citizen involvement in HTA. Evaluation and documentation of good practices can improve these processes, not only in Latin America but also globally.
3. Agencies involved in ETS, including regulators and payers, should engage in discussions with industry and drug manufacturers early in the drug development process to align expectations and objectives.
4. HTAs should include macro and micro levels of evaluation, not only in terms of health outcomes, but also with attention to the evaluation of socioeconomic impact, including quality of life, indirect costs and resources needed to care for patients. These should be mandatory criteria for decision-makers, in addition to the budgetary impact of health technologies. This will require changes in public policies and legislation.
5. Post-marketing research and patient follow-up and monitoring are critical to obtain data because uncertainties and ambiguities may arise due to patient sample size. Agencies should clarify uncertainties early in the development process and generate complementary data to assess the benefits and risks of therapy on an ongoing basis. Industries should commit to providing follow-up data and sharing the long-term risks of therapy.
6. The difficulty of withdrawing a product after approval highlights the importance of differential methodologies and the generation of ongoing data to demonstrate positive results. Agencies must have the ability to change decisions based on new evidence and demonstrate that the therapy remains effective. It is essential to change the decision if the clinical data from the study are not confirmed and the therapy is no longer effective.
7. Leadership is key to effective collaboration among agencies, patients, and industry when it comes to differential HTA for RD.

Relevant information

- Presentation : The role of Patient Organizations in Value/HTA Discussions
- Presentation: Health Technology Assessment in Brazil
- Web Page: GAO Releases Study on the Cost of Undiagnosed and Untreated RD

See complete panel 10 HERE

11. Innovative financing
access and sustainability mechanisms – for comprehensive RD care

PANELISTS

- Dr. Manuel Espinoza
  Head of the Health Technology Evaluation Unit of the Clinical Research Center UC
- Durhane Wong-Rieger
  President and CEO of the Canadian Organization for RD
- Dr. Enrique Teran
  Pharmacologist and President of Sciences of the Academy of Ecuador,
- Angela Chaves Restrepo
  Patient Advocacy Senior Manager Region Intercontinental - Biogen

MODERATORS

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

INTRODUCTION

This panel focused on exploring innovative financing mechanisms, access and sustainable solutions to address the unique challenges faced by governments and nations in providing access to diagnosis and treatment for people with RD in Latin America, including gene therapy. This panel brought together experts in the field to discuss new and innovative approaches to financing and value-based contracting that can be applied to RD, as well as strategies to ensure the long-term sustainability of these efforts.

GENERAL CONCLUSIONS

RDs and orphan drugs present a major financing challenge for health systems that requires a differential and innovative approach. Systems must alleviate the uncertainty inherent to orphan drugs, gradually increase coverage and provide equity. Therefore, it is essential to have a thorough understanding of the behavior of RD, the type of studies used, registration mechanisms, what incentives are appropriate, how to carry out HTA, coverage and financing.

The first thing to consider in financing RD technologies is, what is to be purchased? This takes into account the discussion of the previous panel on HTA assessment that would involve having a value framework and a technology assessment system that allows for the proper characterization of that value framework. Among the options for obtaining the resources needed to fund treatments for people with RDs are single RD funds, for which a value framework is defined solely for RDs. However, if resources come from a general health system fund, a value framework is required that allows different technologies to be valued for both RDs and other diseases. Consideration should be given to the impact of the source of the resources such as an exclusive RD fund or insurance and reinsurance schemes that may dilute the risk and do not go hand in hand with the general health budget.

With this in mind, the approach to RD financing should be prospective. In other words, a long-term view should be taken to identify emerging technologies that are likely to produce the greatest economic and social benefits and thus achieve adequate planning for resource allocation. At the time of purchasing treatments, smart ways of executing the acquisition should be considered.

The first major problem when characterizing value is uncertainty, because even the best scientific evidence comes with levels of uncertainty and uncertainty implies financial risk and risk associated with the value to be produced. This uncertainty can be managed through certain mechanisms. The first type of mechanism requires spaces for negotiation and interaction between the interested parties, which is not found within the legal frameworks of some countries. Having this negotiation space enabled, different mechanisms can be considered, such as prompt payment to the industry, negotiating contracts over a longer period of time and thus obtaining a better price, receiving free units of the product (if permitted by the country's jurisdiction) or negotiating on the basis of portfolios. All these alternatives vary from country to country.

In case the negotiation mechanism is not possible, there are other mechanisms consisting of Risk Sharing Agreements. Among these are two types of agreement, those that manage purely financial risk and those that manage financial risk associated with the production of value. The financial risk mechanisms include caps (if I budget for 10 patients this year and the following year I have 11, the provider covers the eleventh patient); subscription mechanisms, which were implemented in Uruguay (the purchaser pays a subscription for a defined portfolio over time). These schemes guarantee that the resource manager does not spend more than budgeted.

Among the mechanisms that manage financial risk associated with value are payment by results agreements (payment is conditioned on a predetermined outcome); and coverage by evidence (the technology is covered for a limited time with

a specific requirement for the generation and presentation of additional evidence). In Argentina, this type of value-based agreement was implemented to provide coverage for the first gene therapy in the region.

The constant response of decision makers to the request for differentiated models is the lack of resources, but the recent COVID-19 pandemic showed us that resources can be allocated, even without having been budgeted. Challenges also exist in the area of enforcement of regulatory models. Laws on RD have been created in some of the countries, but their implementation is limited by the capacities of the health systems, so that patients resort to seeking treatment through the courts. These cases, far from guaranteeing access, jeopardize the sustainability of the health system. Although many replicable models already exist, thinking about differentiated models can give rise to models of their own for financing RD in the region.

KEY MESSAGES

1. The development of science will continue to drive innovation in RE at an accelerating pace, and governments and societies must be prepared to address the challenges involved.
2. The financing of RD and orphan drugs requires innovative and differential approaches that take into account the inherent uncertainty of these drugs, gradually increase coverage and provide equity.
3. Forward planning is required to identify emerging technologies that are likely to produce the greatest economic and social benefits in order to allocate resources effectively.
4. Uncertainty in value characterization is a major challenge in RE financing and can be managed through innovative financing mechanisms including negotiation mechanisms and risk-sharing arrangements.
5. Although there are isolated efforts that have implemented innovative financing mechanisms for RD in Latin America, in order for them to become more common and sustainable over time, articulation of the different actors is required. Specifically, this requires governments with the political will, structure, competence and technical strength to carry out these efforts; public policies that support and enable this type of mechanism; a pharmaceutical industry that can present this type of proposal; information, care and reimbursement infrastructure; and articulation of the agencies.
6. Access must go hand in hand with timeliness, as delays in access lead to poorer outcomes and higher costs.

7. For health systems to move towards financial sustainability, disease management must be optimized in order to optimize the use of the system's overall financial resources.

Relevant information

- Web page: Introduction of the first gene therapy in the Argentine health system.

See complete panel 11 HERE

12. Ensuring Access to Innovation for Patients with RD

PANELISTS

- Dr. Ignacio Zarante
  President of the Colombian Association of Medical Geneticists, Professor at the Institute of Human Genetics - Pontificia Universidad Javeriana
- Dr. German Escobar
  Former Vice Minister of Health and Protection in Colombia
- Dr. Leonardo Arregoces
  Former Director of Medicines and Health Technology of the Ministry of Health and Social Protection, Colombia
- Dr. Mario Gomez
  Managing Partner, Prospectiva Consulting
- Dr. Natalia Messina
  Director of Special Medicines and High Price in Ministry of Health Argentina
- Dr. Ana Cristina Ochoa
  Medical Director of Takeda.

MODERATORS

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

INTRODUCTION

The central theme was Innovation, Technology and the approval processes of these as treatments in general. With experts from different countries of the region, and different industries, which allowed us to have a vision from various angles of the same problem.

GENERAL CONCLUSIONS

What evaluation agencies recognize as "access" has changed over time. In the beginning, access was considered to be what we know today as coverage, so that decision-makers focused on "allowing" a certain treatment to be used, without considering everything that has to happen for the treatment to actually reach those who need it. Access to innovation for RDs is a challenge in part because of the focus of

health systems on prevalent diseases. Orphan drugs are often associated with the perception of high costs, which is not always true. Patients with RD require an interdisciplinary approach to treatment and care, and access to technology is not the only issue. Access is more than having coverage or insurance. The concept of access has evolved to consider not only drug availability but also the chain of demand, need, and use. Therefore, it is not only a matter of having the drug or technology available in the country, but also of ensuring that all patients who need it can use it under the best possible conditions.

To measure access, indicators must be developed that go beyond whether the drug is available and are based on whether patients with a particular RD have been identified, are using the drug, and have access to all the other care and support they need. Indicators of access and use are available at different levels, such as the national or service provider level. To ensure access to innovation for RDs, it is critical to focus on the entire continuum of care to ensure that patients and their families receive the best possible treatment and support. Currently, with the UN RD resolution, the call is to ensure access, which means working on all the conditions that should enable it, such as the preparation of specialists, referral centers that can treat these diseases, and not least, diagnosis.

Access to innovation for RD varies among Latin American countries. Panelists highlight that some countries still face challenges related to drug procurement and supply due to budgetary pressures and lack of infrastructure. Other countries have achieved a higher level of coverage and procurement of health technologies, but face difficulties in accessing quality technologies in a timely and effective manner. Cross-sectoral support is critical for patients with RD, their families and caregivers. Some countries have achieved the first two stages of access and are now focusing on intersectoral support and social protection for patients with RD and their families.

In Colombia, access to technology for RD is a major challenge, and the current government's position on access to innovative technology is unknown. Despite the development of public policies and advances in this field, access to treatments occurs mostly through legal actions (tutelas). In fact, most of the legal actions in the country are for orphan drugs, representing 70% of the reimbursements in the country, with 92% of these treatments reimbursed last year. The panelists agree across all countries that legalization of treatments for RDs is not an effective solution to ensure access to treatments. While it may be the only option for some patients, the process of going through the justice system to obtain a drug or therapy can be time-consuming and costly. It is a costly process for both the patient and the health system, as it may cost the system more to pay for a drug or therapy for an individual patient at market value. In addition, the judicialization of treatments for RDs puts health care decisions in the

hands of the legal system rather than health care providers. The health care system should be responsible for solving the problem of access to RD treatments, not the legal system.

Measuring the therapeutic value of health technologies for RD is complex, as the value is not necessarily assessed with traditional survival variables but with quality of life measures, functional scales, and other metrics. This complexity echoes previous calls for a more sophisticated approach to measuring the value of health technologies for RDs and ensuring access to these technologies for those who need them.

KEY MESSAGES

1. Access to treatment for RDs is more than just having coverage or insurance; it involves ensuring that patients can actually access and use the treatment they need in a timely manner.
2. To ensure access to innovation for RDs, it is critical to focus on the full continuum of care to ensure that patients and their families receive the best possible treatment and support.
3. To improve timely access, it is critical to educate primary care physicians, pediatricians, and gynecologists in neonatal screening and to recognize the signs and symptoms of RDs and appropriate referral or referral situations. By providing appropriate training to primary care physicians, patients can receive timely diagnosis and prevention, which can help reduce the cost of treatment and have a significant impact on the overall healthcare system.
4. Legalization of treatment or access to treatment is sometimes the only option for some patients, but it is not effective in providing timely and adequate treatment.
5. It is the collective responsibility of all stakeholders, including governments, the pharmaceutical industry and other organizations, to work together to find solutions to the challenges posed by RDs.

Relevant information

- Report: CONETEC Report on the Evaluation of Treatment (gene therapy) in Argentina

See complete panel 12 HERE

13. The Digital Revolution
Digital health, Information technology and Artificial Intelligence in RE

PANELISTS

- **Dr. Elmira Safarova**
  Co-Founder and CEO Rarus Health SPA
- **Dr. Marisa Aizenberg**
  Academic Director of the Health Observatory of the Faculty of Law, University of Buenos Aires (UBA)
- **Dr. Luis Pino**
  Medical Oncologist, Founder and CEO of OxLER
- **Irene Kanter-Schlifke**
  Director of Strategy and Customer Information, Biogen Digital Health

MODERATORS

- **Felicitas Colombo**
  Director of Government Affairs AHF
- **Dr. Mariana Rico**
  Medical Director of AHF

INTRODUCTION

Differentiating between digital health and artificial intelligence (AI) is essential to understanding their role in RDs. Digital health refers to the integration of various technologies to optimize health, while AI involves computational cognitive processes that mimic human thinking and is a key component of digital health. Understanding this distinction is crucial, as the two are complementary but distinct elements in the context of RDs and orphan diseases.

GENERAL CONCLUSIONS

RDs pose challenges due to lack of awareness and knowledge among healthcare professionals, highlighting the need to incorporate AI into medical decision making. AI can aid in the prediction and classification of RDs, addressing diagnostic limitations and disease progression. Predicting associations between genetic profiles, phenotypes and RDs, as well as classifying and making predictions of progressive disease, are areas where AI and machine learning techniques, such as deep learning, can contribute significantly. These advances have the potential to overcome the limitations of human cognition and improve disease diagnosis and understanding.

The intersection between AI and digital health includes automation and software applications that help patients. These technologies should be treated as medical devices, evaluated and integrated into the comprehensive treatment of patients with RD to ensure effective patient follow-up, data capture and generation of real-world evidence. Treating them as medical devices ensures effective patient monitoring, capturing essential patient data and generating automatic real-world records, which can further aid in the research, treatment and advancement of RD management.

The lack of data in RD poses a major challenge for research and therapy development. To address this, digital tools are being developed to empower patients and contribute to drug development. However, there are challenges in integrating technology into healthcare systems, as there is a delicate balance between traditional and digital approaches that need to be taken into account. Patient involvement in decision making and data collection is crucial. By providing tools such as symptom questionnaires, patients can actively contribute to their own healthcare and provide valuable information for healthcare professionals such as pediatricians.

Electronic health records are also important for real-world evidence and clinical practice, but the lack of unified standards and the limited number of patients in RD make data management a challenge. The need for a centralized and standardized approach to managing electronic health records and patient information is essential in RE. Currently, different clinics have their own standards, resulting in varied and fragmented data.

The region still faces a major challenge in the transition from paper records to digital systems. The process of digitizing medical information raises concerns about privacy, legal issues and the overall management of digital healthcare data. The panel emphasized the potential of healthcare digitization as a solution. It presents an opportunity for improved access to healthcare services and improved public policy. The implementation of telecare programs in countries or even in the region can help overcome the geographical distribution of specialists, reducing the time needed for diagnosis and improving overall healthcare outcomes.

The panel highlighted the importance of a patient-centered approach to digital health initiatives. Patients should be involved in the design and development process to ensure that digital tools and technologies meet their needs. The panel highlighted the importance of ethical considerations and data protection in the digital revolution. With the rise of technologies such as AI and precision medicine, it is crucial to have a thorough ethical understanding of how data is collected, stored and used. Strong frameworks and regulations are needed to ensure the privacy and security of medical information, avoiding potential harm and discrimination caused by the misuse of data.

Citizen awareness and participation are crucial in shaping these frameworks and demanding a safe and appropriate digital health environment.

KEY MESSAGES

1. It is important to differentiate the roles of digital health and AI in RDs. Digital health integrates technologies for health optimization, while AI involves cognitive processes and is a key part of digital health.
2. AI and digital health software supporting patients and automation should be treated as medical devices. They should be evaluated and integrated into comprehensive RE treatment for effective patient monitoring, data capture, and real-world evidence.
3. Digital tools address access and policy issues, especially in regions that rely on paper-based systems. Telecare and other digital solutions can improve access to care, particularly in underserved areas.
4. Digital tools should have a patient-centered approach that prioritizes patients and their needs, involving them in the design process. A regulatory framework should be developed or strengthened to address ethical and legal concerns, including data privacy, to prevent exploitation and discrimination.
5. Precision medicine and AI offer improved diagnostic and treatment outcomes, but require ethical considerations and proper data management. Secure environments and risk management plans must be implemented to protect vulnerable systems.

Additional information

- App/Website: Physiotherapy for People living with neuromuscular diseases (Biogen) https://portal.physio.me/login
- Web Page: Postgraduate in Digital Health University of Buenos Aires
- Website: OxLER
- Website: Rarus Health

See complete panel 13 HERE

14. Advancing clinical trials and RD research in Latin America – Diversity and inclusion in genomics research

PANELISTS

- **Dr. Gabriela Repetto**
  Director of the RD Program, Faculty of Medicine, Clínica Alemana Universidad del Desarrollo,
- **Dr. Roberto Giugliani**
  Co-President of Casa dos Raros
- **Dr. Deborah Requesens**
  Director of Jumpstart Program, Orphan Disease Center, University of Pennsylvania
- **Dr. Reggie Garcia Robles**
  Medical Geneticist and Salubrista, Research Professor, Pontifica Universidad Javeriana

MODERATORS

- **Felicitas Colombo**
  Director of Government Affairs AHF
- **Dr. Mariana Rico**
  Medical Director of AHF

INTRODUCTION

This panel focused on the current status and future developments of clinical trials and RD research in Latin America. The panel featured experts who shared their knowledge and experience on the design and implementation of RD clinical trials, the challenges and barriers to conducting research in the region, and the importance of diversity and inclusion in genomics research. The panel had discussions on the potential benefits of expanding RD research in Latin America, including improved diagnosis, treatment and care for patients and families affected by these diseases. The panel provided a valuable opportunity for attendees to learn about the latest advances in RD research and to discuss the challenges and opportunities for improving diversity and inclusion in clinical trials and RD research in the region.

GENERAL CONCLUSIONS

Conducting clinical trials in Latin America for patients with RD can generate early access to new therapies that would otherwise take many years to become available. Clinical research raises the standards and improves the quality of healthcare services provided by participating centers. Panelists emphasized the importance of studying the genetic characteristics of Latin American populations to improve molecular diagnostics and develop targeted treatments. By obtaining information on specific genetic variations and alterations, researchers and healthcare professionals can improve precision medicine approaches and tailor treatments to the unique genetic characteristics of patients in the region. Research into population-specific diseases and their characteristics can guide effective strategies. This is supported by examples such as variations of Huntington's disease found in the region and certain conditions influenced by ancestral indigenous components. By studying these population-specific diseases, researchers can gain a deeper understanding of their origins, prevalence and clinical characteristics, leading to better planning and the development of effective strategies for diagnosis, treatment and prevention.

The population and genetic diversity of Latin America makes it a favorable location for clinical trials. Panelists noted that the population of Latin America, particularly in urban centers, offers opportunities for clinical research due to its proximity to research centers and a significant number of RD cases. The region's genetic variations and distinct genetic profiles also present potential for studying the efficacy of existing drugs and developing new therapeutic approaches.

However, there are challenges to conducting clinical trials in Latin America, including lack of necessary infrastructure, regulatory inefficiencies and long evaluation timelines. Managing regulatory timelines and aligning clinical trial evaluation processes for RD clinical trials are crucial to include Latin American patients. Overcoming administrative hurdles and ensuring free access to investigational drugs during trials helps to attract more studies and advance research efforts. Improvements have been made in Brazil, such as simultaneous ethical and regulatory evaluations and an accelerated process for RD. Such regulatory advances have allowed more studies to be conducted, including the elimination of a regulation that required drugs to be administered to patients throughout their lives. Lengthy evaluation timelines and sequential evaluations may delay patient inclusion in trials, as patients from developed countries may fill available slots. Improving regulatory frameworks, including simultaneous evaluations by medical and ethical agencies, can create a fast track for RDs, prioritize patient inclusion, and attract more studies to the region.

In Chile, the Ricarte Soto Law has provided coverage for many patients with RD, but is restrictive for research and clinical trials. These limitations include the provision

of investigational drugs for the lifetime of the patient, which poses challenges when a drug is found to be ineffective. Indemnification requirements and additional criteria for conducting research in people with intellectual or cognitive disabilities also create barriers to trial participation.

The panel recognizes the historic achievement of obtaining approval for the first gene therapy clinical trial in Latin America. While recognizing that progress has been slower compared to other regions, this approval represents a crucial step in the advancement of clinical trials for RD in the region. The Pan American Health Organization (PAHO) prepared a document highlighting the regulation of advanced therapy products for therapeutic purposes. It mentions the collaboration and contributions of the regulatory authorities of countries such as Brazil, Canada, the United States, Colombia, Chile, Ecuador, Mexico and Cuba. In this rapidly changing field, it is important to keep up to date with regulatory developments and there is a clear need for continued cooperation and networking among countries.

One of the challenges to be addressed is health literacy: raising public awareness about the development of scientific knowledge and research and dispelling misconceptions and resistance to participating in clinical trials due to concerns about experimental procedures or potential risks. Educating the public and creating a scientific culture is necessary to counter resistance to scientific knowledge. This was demonstrated during the pandemic through the contrast between global confidence in scientific knowledge and resistance to scientific understanding.

Underrepresentation of diverse populations in clinical trials and research is a major problem in Latin America and the United States. In the U.S., despite being 19% of the population, minority communities are underrepresented, with only 4-6% participating in clinical trials and less than 1% participating in NIH-funded research. The lack of diversity in genomic research and clinical trials is influenced by several factors, including language barriers, cultural differences, lower socioeconomic status, limited access to genetic testing, and concerns about privacy and trust. These factors contribute to the lack of diagnosis and limited participation in health systems that incorporate advanced technologies. Efforts are underway to close the diversity gap in research and clinical trials. Initiatives include translating consent documents into different languages, addressing cultural nuances, promoting representation in leadership roles, building trust through community engagement, and educating future healthcare professionals about RD and the importance of diversity. Collaborative efforts between institutions, researchers, and minority communities aim to increase representation and improve understanding and treatment of RD.

Collaborations between public and private healthcare institutions can help overcome the challenges in conducting clinical trials for RD in Latin America. While

private hospitals may have the necessary infrastructure, research teams and expertise are often located in public hospitals and universities. Finding the ideal combination of skilled teams and adequate infrastructure and human resources can be a major challenge, but it is crucial to the success of clinical trials. There is a need to strengthen collaboration and networking among investigators across national borders. Improving cooperation and harmonizing regulations would facilitate clinical trial initiatives, which can lead to more efficient and effective research outcomes. In addition, efforts are being made to transfer knowledge and technology from institutions such as the University of Pennsylvania to Latin American countries. This transfer is aimed at facilitating clinical trials by sharing experience, regulatory knowledge and manufacturing capabilities.

KEY MESSAGES

1. Conducting clinical trials in Latin America can generate early access to new therapies for patients with RD, and reduce the time it takes for these treatments to become available to the general population.
2. Studying the genetic profiles of Latin American populations is crucial for improving molecular diagnostics, developing targeted treatments, and improving precision medicine approaches. Understanding specific genetic variations and alterations allows for personalized treatments that take into account the unique genetic characteristics of patients in the region.
3. The population and genetic diversity of Latin America makes it a favorable place to conduct clinical trials for RD.
4. Challenges to conducting clinical trials in Latin America include infrastructure requirements, regulatory inefficiencies and long evaluation timelines. However, advances have been made in countries such as Brazil, such as simultaneous ethical and regulatory evaluations and an accelerated process for orphan diseases. These advances speed up the approval of clinical trials and attract more studies to the region.
5. Addressing health literacy and increasing diversity representation in clinical trials are important challenges. Raising public awareness of scientific knowledge and research, as well as closing the diversity gap in research and clinical trials, are crucial to advancing RD treatments in Latin America.
6. Collaboration between public and private health institutions, as well as networking among researchers across national borders, can help overcome challenges and facilitate clinical trial initiatives in Latin America. Networking, sharing expertise and regulatory knowledge, and promoting knowledge and technology transfer can improve research outcomes and efficiency.

Additional information

- Publication: The regulation of advanced therapy products for therapeutic purposes.
- Presentation: Social Appropriation of Science, Technology and Innovation (Spanish)
- Website: Orphan Disease Center, University of Pennsylvania

See complete panel 14 HERE

15. Integrating RE Ecosystem Stakeholders
Regional engagements

PANELISTS

- **Diego Gil**
  President of the Colombian Federation of RD - FECOER
- **Dr. Claudia Gonzaga Jauregui**
  International Laboratory for Human Genome Research, UNAM
- **Dick Salvatierra**
  Founder and CEO Americas Health Foundation
- **Yaneth Giha**
  Executive Director, FIFARMA
- **Dr. Gustavo Mendes Santos**
  Former General Director of Medicines and Biological Products at the National Agency of Health Surveillance, ANVISA
- **Dr. Ignacio Zarante**
  MD. President of the Colombian Association of Medical Geneticists - Professor at the Institute of Human Genetics - Pontificia Universidad Javeriana
- **Heidi Bjornson-Pennell**
  Chan Zuckerberg Initiative

MODERATORS

- **Felicitas Colombo**
  Director of Government Affairs AHF
- **Dr. Mariana Rico**
  Medical Director of AHF

INTRODUCTION

To translate discussions into action, a list of priorities must be created jointly, including the pharmaceutical industry, governments, regulators, patient organizations, geneticists and physicians, as the joint impact is more noticeable. Coordination of activities maximizes the collective impact of investments in RD research, creating a more effective dialogue, a registry with common databases, improved innovative therapeutic approaches and increased information and access to patients.

GENERAL CONCLUSIONS

There is much work to be done for the benefit of patients with RD. Opportunities have been identified, including the collaboration and participation of all actors in the creation of spaces, platforms, and proposals to improve public policies not only in the health but also in the social area. Priorities include investment in the generation of follow-up data on the effectiveness of therapies, since there are currently no mechanisms to replace therapies that are no longer effective. It is also important to build bridges of collaboration for the participation of international patient organizations in the region, which can contribute with knowledge and technology transfer, and in turn, learn from local practices that can favor their intervention at the global level.

Heidi stresses the importance of collaboration and philanthropy in supporting RDs. She emphasizes the need to connect patient communities across borders and highlights the role of patients in shaping research priorities. She encourages philanthropic and funding organizations to continue to support patient organizations and research efforts in the region. She also highlights the international nature of RDs and the formation of a community dedicated to addressing them.

Dr. Mendes stresses the importance of identifying priorities and undertaking collective actions to achieve significant results in the field of RD. He emphasizes the need to invest in data generation and monitoring to ensure reliable results and adaptability in therapies and diagnostics. In addition, he highlights the crucial role of patient involvement in decision-making processes. The work is ultimately for the benefit of patients and their involvement is critical.

Diego highlights the importance of collaborative networking and inclusive engagement among stakeholders to address RE in Latin America. He highlights the need for synergy and harmonization among ecosystem actors, recognizing that this is a challenging task. In his experience as a patient advocate, he echoes the challenges of getting a diagnosis, accessing specialized care and treatment. Despite the significant barriers, it is crucial to work together and not navigate the journey alone. Diego emphasized the value of this Congress in shaping public policy and fostering social cohesion. He expressed admiration for the leaders of patient organizations, particularly those working at the local level, who effectively connect different stakeholders and work for policy implementation. The need for continued progress in the region through collaborative efforts is evident.

Dr. Zarante describes the transformative experience of attending a congress and emphasizes the importance of translating the knowledge and connections gained into actionable results. He stresses the importance of networking and staying connected with the individuals and patient groups that met during the congress.

There is a need for action and reference examples from other countries as motivation to implement similar initiatives. Working in groups and involving all stakeholders, including the pharmaceutical industry, government, regulators, patient organizations, medical professionals and academia, is crucial to achieve meaningful progress. The key message is to harness the potential impact of collective action: foster collaboration, leverage collective expertise and engage with all stakeholders to drive positive change in the DR landscape.

Yaneth highlights FIFARMA's role in conducting studies and collaborating with Latin American actors to identify tasks and take actions in the field of RD. She emphasizes the importance of building capacity and leveraging connections to bring in experts who can contribute to improving access to innovative treatments and contracting mechanisms. Yaneth also emphasizes the continued need for education and awareness and encourages persistent efforts to advocate for RD, stating that the task requires long-term commitment from all stakeholders, including industry. Persistence and repetition are crucial to delivering a message effectively. When you feel exhausted from repeating something, it is a sign that people are starting to understand and listen. Therefore, in the context of RD, it is important to persist and

keep repeating the message, as it may take years or even decades of constant effort to achieve the desired result.

Dr. Gonzaga-Jauregui emphasizes that access and collaboration are crucial to address the challenges faced by people with RD in Latin America. Access to health services, particularly for those with low socioeconomic status and in marginalized communities, is essential to ensure equity and inclusion. While there are additional challenges related to access to treatment and regulation, it is important to prioritize and make efforts to reach remote and underserved communities, ensuring that technologies and diagnostic services are available to all people with RD. Achieving equity and inclusion is necessary to benefit all patients with RD.

Dick Salvatierra discusses the importance of collaboration among diverse stakeholders in the RE field. Collaboration with academic research institutions, companies, regulatory bodies, patient advocacy organizations and international associations can maximize the collective impact of RD investments. This collaboration can accelerate progress in areas such as faster dialogue, establishment of common registries and databases, innovative therapeutic approaches, and increased patient information and access. It highlights the need for resources to support collaborative efforts and expresses gratitude to the various groups and organizations that made the Congress possible. The message emphasizes that collaboration is crucial for future efforts and invites continued collaboration for future congresses and initiatives.

Additional information

- Publication: Access Survey on availability and access to innovative therapies , FIFARMA.

See complete panel 15 HERE

VI. **Comments**

from RD Patient Organizations

Chile

Regarding attendance and invitation to discuss RD issues, prior to the Congress we were already working on an agenda with the Ministry of Health and the Congressional Commissions in both chambers. However, after the Congress and the participation of influential actors in our country (Dr. Repetto, Dr. Daza and Dr. Espinosa) we have noticed a more coordinated intention in the plans presented to our authorities. In addition to this, our RE Law project underwent changes in its articles without consulting the communities, which caused a tense situation that, thanks to informal conversations during the Congress, could be resolved or redirected. Regarding new stakeholders, we have established links since the end of the Congress, some of them are starting research in our country and others who had been working in Chile for some time and had no knowledge of our area of performance and company. As for new work agendas, so far we have consolidated a community of Neurofibromatosis and Rarus. Now, regarding traffic on social networks, according to META data, the publications had a reach of 3,014 people on Facebook and 1,079 on Instagram, 72% less than our common publications on Facebook, but in this case it was organically (without promotion payment). The median number of reactions was 17, and 116% more messages arrived during the publication of news about the Congress. Finally, we consider important to mention that the organization of the Congress was wonderful and opens the appetite to want to repeat the experience and make it a good habit, however it is necessary to add other aspects of work and development, which allow greater visibility to the effects and impact that this platform has - will have in the region.

Dominican Republic

Regarding attendance and invitations to discuss RE issues, we are currently continuing our efforts with the Senator of the National District, Faride Raful, for the drafting of the RE bill. For this purpose, we are still in the process of confirming the work tables. Regarding approaches with new stakeholders, during the congress we had several approaches with regional patient associations that were interested in putting us in contact with patients in our country that are not yet grouped. For this, we are already in contact with Vicky Arteaga from the Syngap Research fund, who put us in contact with two mothers of Syngap 1 patients. As for new work agendas, we are coordinating a working session sponsored

by a laboratory, where we will include the main stakeholders of the country to shape the whole issue of the bill. This was coordinated during the congress. Now, in terms of social media traffic, we had some interactions during the congress. Several people from regional funds in the Dominican Republic approached the ADAPA leader to include their patients in the umbrella of the RD network in the Dominican Republic. Regarding new approaches with the industry, there was an approach with Sanofi. A meeting has been scheduled with the laboratory and with Dr. Siu Chang. This link was created thanks to the congress because Sanofi representatives approached them to coordinate a meeting and thus give shape to a full day of work with stakeholders to seek the creation of the RD law in the Dominican Republic. Doctors, ADAPA foundation staff, Sanofi industry and decision makers from the political class will attend. Finally, it is important to add that the congress definitely opened doors for us and put us in the regional context, it has given us a lot of visibility. We now have a broader knowledge of the reality regarding RD in the whole region. Thank you very much for allowing us to be part of such an important and successful event.

Panama

As for new work agendas created as a result of the congress, we are about to arrange meetings with 2 health entities. As a Federation we consider that the congress gave us a significant formation and impulse as a group, we congratulate you for the professionalism and height with which the congress was carried out. Sixty percent of the members of the alliance were connected to the event and affirm that they would participate again, that the content and management of the agenda was satisfactory.

Mexico

The perception of the presentations, opinions and suggestions were of great importance, very important notes were taken and I also have as regional referents all those who participated. New stakeholders were approached and this is considered a resource of great value since the exchange is decisive in strengthening the network of support and knowledge that exists in the region. On the other hand, we are in the construction of new meetings and joint projects, this activity is what gives visibility to

the group and shows the strength of the associative movement. As for the traffic in networks, these increased especially during the transmissions during the event, which increased the conversation of patients with topics related to the event itself. It is of great importance to follow up and build projects that give continuity to the topics discussed, because thanks to the congress, expectations have been opened.

Peru 🇵🇪

Regarding the approaches to the public and private sector, in the public sector, a summary concept note and priority points of conclusions are required to reinforce the developed topics and consensus, to be sent to the public and private sector. In addition to this, there were requests for meetings for FEPER from 3 companies in the industry. We will initiate activity with one of them in June. The congress allowed us to link 2 patient associations with the industry for specific pathologies and they will have working meetings. As a recommendation for future events, more industry representatives in booths and more patient associations in booths or modules that allow individual dialogues. Thanks to the congress, there was a linkage with regional expert stakeholders in various fields. In addition, new projects were created for FEPER, mainly related to ultra-RD or diseases that do not have a patient association in our country. They will start in June 2023. Approaches have arisen mainly with the industry, a new one for FEPER and reinforcement of pre-existing ties with three. ERCAL's image has been positioned with a strong impact in Peru and all Latin America. We also consider that there are possibilities for the growth of FEPER and other specific associations to diversify funding and expert actors in the field at regional level, possible forums or dialogues on aspects that Peru must improve. As a result of the congress, much expectation was generated in patient associations, patients, among others. So much so that they continue to tag #ERCAL in individual publications. We believe that it was necessary to publish the event live on the participants' websites for it to have more impact. In addition, some patients and leaders reported not knowing how to use streaming. We believe that it should be published via Facebook or YouTube to generate more traffic to the entire live event.

Ecuador

In terms of attendance or official invitations to discuss RE issues, we have participated in two very important events related to RE. Our line of work is public policy and we continue to open spaces for dialogue with the national government. Regarding approaches with new stakeholders, we have achieved some approaches to some people who are developing in this field. In addition, based on the RE congress, we have broadened our scope, and we have had dialogues with several colleagues to develop projects together. Regarding approaches with the industry and in accordance with the RD congress, we had conversations with several representatives of the pharmaceutical industry, and we have continued to have virtual chats to present projects in favor of patients with RD. Being an organization in full development, and having been invited to the congress, the expectations of our associates have grown and we consider that it has been a valuable point to be part of this group of organizations of international stature. We hope that we will be able to develop a regional project aimed at early detection and care of RD. Based on neonatal screening, karyotypes and genetic studies and thus educate and educate all of us who are involved in this issue of RD in the region.

Colombia 🇨🇴

The congress held in Bogota continued to position FECOER in the field of RD at the international level. We received invitations to be speakers at the following international events: World Patients Congress. Geneva, Switzerland. May 19 and 20, 2023 organized by the International Alliance of Patients Organizations (IAPO), Global Patient Partnership Summit. Miami, USA. May 22, 23 and 24, 2023 organized by Boehringer Ingelheim, World Orphan Drug Congress 2023. Washington DC, USA. May 25, 2023 organized by WODC USA / Terrapin, ER Scenario. São Paulo, Brazil. June 16, 2023 organized by FEBRARAS / Casa Hunter. In addition to this, we connected with María Belén Jaimes, new referent of Orphan / RD of the Ministry of Health and Social Protection. Added to this, there was an increase in the number of RD patients, caregivers and families who have contacted FECOER requesting information about their diagnoses, medical guidance, legal guidance or because they wish to connect with support networks in the community. We worked collaboratively with Elmira Safarova on webinars on the use of data to improve access to innovative therapies. With Dr. Ignacio Zarante, the Colombian Association of Medical Geneticists, the San Ignacio University Hospital and the Pontificia Universidad Javeriana, we will evaluate the implementation of the Casa dos Raros project in Brazil and its possible adaptation to Colombia to promote the establishment of reference centers. Approaches were made with: Optimal Therapies, Raras CRO, Quindio Biotech and we strengthened the relationship with other laboratories with which we already have contact. As areas of visualization of growth are the relationship with LATAM federations, the relationship with opinion leaders at scientific level and the knowledge of the regional panorama.

We conducted feedback with federated leaders who highlighted the following; in terms of positive aspects, a comprehensive and technical academic agenda was presented with very relevant opinion leaders on a regional and global scale, there was a very important collaboration between AHF, patient groups, academia, scientific societies and the pharmaceutical industry. The AHF staff did an excellent job of coordination and management, creating integration among stakeholders and opening new opportunities for collaboration. The positioning of the work of the LATAM patient groups was achieved, integration between the leaders of the LATAM patient groups was created and the logistics and production was impeccable. As for aspects to improve, there was a lack of question and answer sessions and to take into account the accessibility conditions for people with reduced mobility.

Costa Rica

After participating in the First Latin American Congress of RE, we set ourselves the task of coordinating several meetings to inform our members about the scope, improvements and everything that will come within the strategic plan that we have as a Network. So we established three work commissions. Within the work, we are making an approach with the Academy in order to start a project that intends to be a model at local and international level. Through some members who live in remote areas of the country, we are approaching other regional hospitals in order to be able to disseminate material related to RD and the work we do. We are creating youth spokespersons for the Network, as well as a Latin American Seminar together with one of our member associations and the Ecuadorian Federation of RD. We are achieving the support and joint work with the Academy (Universities) and as a result of the visibility and interaction we have in our different channels, we are opening possibilities to project in other countries what we will do with the launching of our channel in Youtube. There were approaches with the industry, currently, we manage a cordial relationship with the sector and in fact many seek us to work together with training and others (Bioplus, Stein, Astrazeneca, Sanofi).

VII. Acknowledgments

Partners

ADAPA

alsser
ALIANZA DE ASOCIACIONES DE ENFERMEDADES RARAS HUÉRFANAS Y POCO FRECUENTES DE PANAMÁ

FEBRARARAS

FECHER
FEDERACIÓN CHILENA DE ENFERMEDADES RARAS

FECOER
Federación Colombiana de Enfermedades Raras

feper
Federación Peruana de Enfermedades Raras

FERPOF
Federación Ecuatoriana De Enfermedades Raras O Poco Frecuentes

FIFARMA

omer
Organización Mexicana de Enfermedades Raras

Enfermedades RARAS CR

REMEXER
Red Mexicana de Enfermedades Raras

Prospectiva
PUBLIC AFFAIRS LAT.AM

Sponsors

GOLD

HORIZON

Biogen

Takeda

Chan Zuckerberg Initiative

AHF
AMERICAS HEALTH FOUNDATION

SILVER

illumina

ALEXION
AstraZeneca Rare Disease

RARAS CRO

sanofi

optimal therapies
A DIVISION OF salutis

ultragenyx

Boehringer Ingelheim

Report Made by

Mariana Rico, Sophia Harb and Carlos Martínez Villela.

Design and Edition by José Carlos Parra.

2023

1ST CONGRESS ON Rare Diseases

LATIN AMERICA AND THE CARIBBEAN

AHF AMERICAS HEALTH FOUNDATION

ERCAL ENFERMEDADES RARAS DEL CARIBE Y AMÉRICA LATINA

Chan Zuckerberg Initiative

IHS

Pontificia Universidad JAVERIANA Bogotá

FECOER Federación Colombiana de Enfermedades Raras