

*Final Version eng, July 17, 2024*

# 16<sup>th</sup> ICORD Annual Meeting: “Incentivizing Science and a Comprehensive Program for Rare Diseases”

Venue:

“Sala de Conferencias”, main level, Faculty of Pharmacy and Biochemistry, University of Buenos Aires (UBA). Junín 956, Autonomous City of Buenos Aires (CABA), Argentina.

Date:

July 24 and 25, 2024

Hosts:

**ICORD**, International Collaboration for Rare Diseases and Orphan Drugs ([www.icord.es](http://www.icord.es))

**Faculty of Pharmacy and Biochemistry- University of Buenos Aires**, ([www.ffyb.uba.ar](http://www.ffyb.uba.ar))

**CERYDH**, (Delegate for ICORD Congress at Argentina), Colaboración para las Enfermedades Raras y Drogas Huérfanas, ([www.cerydh.com.ar](http://www.cerydh.com.ar))

Institutional Partners:

**IRDIRC**, International Rare Diseases Research Consortium

**RIBERSER**, Red Iberoamericana de Enfermedades Raras

**CONICeT**, Consejo Nacional de Investigaciones Científicas y Técnicas.

**IORO**, Indian Organization for Rare Diseases

**CORD**, Chinese organization for Rare Diseases

**CERYD**, Centro de Referencia en Enfermedades Raras y de Dificultoso Diagnóstico.

**Latiner SA**, Enfermedades Raras Latinoamérica.

**FMF**, Fiebre Mediterránea Familiar, Argentina

Media Partners:

**SAPeM** (Sociedad Argentina de Periodismo Médico-AMA), **Periodismo y Salud, Hablemos de Salud.**

Sponsors:

**Qualix DoT**

**Empresa**

**Empresa**

**Empresa**

(Sponsorship of this meeting is devoted to logistic support and not related to the program content).

### Meeting Aims:

1. To promote the implementation of comprehensive health programs for the care of rare diseases and the development of orphan drugs in accordance with international models adapted to Latin American realities.  
It emphasizes the need for programs to prioritize based on the local health situation and be developed in alignment with the social and economic conditions of each country.
2. Suggesting working models and incentives for local scientists and the health industry to generate orphan diagnoses and treatments in collaboration with other countries world-wide and at the highest standard possible.

### Welcoming Remarks

Welcome to the 16th annual meeting of ICORD, this time developed in conjunction with the Faculty of Pharmacy and Biochemistry of the University of Buenos Aires (UBA). This event is an academic effort to gather important information related to lesser-known diseases, rare diseases, and drugs that must be researched and developed under special conditions, orphan drugs.

The issue is of utmost importance for these times in the country and the region, since it is about including in the health care scheme very relegated diseases, difficult to diagnose and with little or no available treatments. Or when there are treatments, they are difficult to access. At the same time, rare diseases are a valuable opportunity for research in a country that has excellent scientists and a pharmaceutical industry installed with export quality. However, it is not yet focused on this topic due to lack of appropriate incentives. So, the bridge with international experts, who bring with them successful models in other countries, is the starting point to begin to change the history of rare diseases and orphan drugs in the country.

ICORD was already in the country in 2010 and then laws, regulations, education and care centers and patient organizations more aware of collective work appeared. Rare diseases became visible.

Now is the time to think that the joint work of scientists, health professionals, patients, government officials and industrialists can work together to generate care models that are sustainable and accessible and research programs that generate innovation projects, translation towards production of high quality and export resources, generating a balance in health spending, just at times when costs increase, and available resources are limited.

All of this can be debated, exchanged experiences, made connections, and put together ideas, in a double academic day that public education offers to its people and the people of the region.

We thank the national and international institutions that support the congress, the communication media and those who with their personal efforts have contributed to making this event possible. We hope that this conference will be inspiring and serve as a true platform for collaboration for all rare diseases.

On behalf of the Organizing Committee.

---

**Program:**

\*On-line presentation

Simultaneous Spanish/English translation service is provided throughout the day on July 24.

**Day 1: July 24, 2024;** Conference Room

On-site registration: 08:00-16:00, secretariat desk.

**08:30-09:15 Session I: Opening 16<sup>th</sup> ICORD Annual Meeting.**

Introduction: Sr. Marcelo Villanueva (Argentina) CERYDH

-Emilio Roldan (Argentina), CERYDH Scientific Coordinator

-Cynthia J. Tifft (USA), President Elected of ICORD

-Daniel Salamone (Argentina), CONICeT President

-Pablo Evelson (Argentina), Dean Fac Pharmacy and Biochemistry (UBA)

**09:15-10:30 Session II: Collaboration for expanding diagnosis and research for rare Diseases.**

Sessions moderator: Emilio Roldan

-María T. Acosta (USA) NIH, The NIH Undiagnosed Diseases Program:  
International Collaborations.

\*Claudio Carta (Italia) ISS, “Buenos FAIRes”: why and how you should apply the FAIR principles to your research project.

-PJ Brooks (USA), DRDRI-NCATS, NIH. Beyond “One Disease at a Time”: Platform Approaches for Rare Disease Gene Therapy/Gene Editing Clinical Trials.

10:30-11:00 break

**Posters Sessions A**, 1<sup>st</sup> floor, Authors of Posters 01 to 07

**11:00-12:15 Session III. Genes Diagnoses and Therapies.**

-David R. Adams (USA), NIH, Genomic Approaches to Undiagnosed Diseases:  
Lessons from International Collaborations.

-Cynthia J. Tifft, (USA), NIH, A phase I/II First-in-human gene therapy trial for GM1 Gangliosidosis: Interim results.

-Mireia del Toro (Spain), Access to CNS rare diseases innovative therapies.

12:15-13:45 Lunch break

12:30 Business lunch (By invitation). Collaboration for rare diseases research. Lecturers room.

**13:45-15:00 Session IV. The strength of international collaboration for rare diseases**

-David Pearce (USA), IRDiRC: About IRDiRC programs and international collaboration

\* Anneliene Jonker (The Netherlands), IRDiRC, Twente University: Pluto Program (living no one behind)

\*Anneliene Jonker (The Netherlands), IRDiRC, Twente University: The relevance of repurposing drugs for rare diseases.

15:00-15:30 Break

**Poster Sessions B**, 1<sup>st</sup> floor, Authors of Posters 08 to 14

15:30-16:15 **Session V Initiatives to reduce the economic impact of Rare Diseases on healthcare budgets.**

\*Alberto Mantovani (Italy) ISS. Primary prevention of Rare Diseases and the new One Health vision.

\*Domenica Taruscio (Italy) ISS. Extended newborn screening for rare diseases: the Italian system.

16:15-17:30 **Session VI: Example of rare diseases achievements in countries with different health realities and resources.**

\*Rumen Stefanov (Bulgaria), The Institute of Rare Diseases of Bulgaria

-Ramaiah Muthyala (India), IORD initiatives at India.

-Mireia del Toro (Spain), Val d'Hebron Hosp. The model for assisting rare diseases developed at Catalunya.

-Luis Alejandro Barrera (Columbia), Univ Javeriana de Bogotá. Rare Diseases initiatives at Columbia

17:30 -Emilio Roldan, Marcelo Villanueva, Summary of the day.

---

**Day 2: July 25, 2024;** Conference Room

On-site registration: 08:00-13:00, secretariat desk.

08:30-10:00 **Session VII Researching in the field of Rare Diseases.**

Coordinators: Patricia Setton, Javier Santos

- Tatiana Guendulain (Argentina), Potential therapeutic use of bacterial flavin monooxygenase to trimethylaminuria.
- Mario Contin (Argentina) Synthesis and supramolecular study of an amphiphilic compound for the vehiculation of idebenone5
- Micaela Carcione (Argentina), Current perspectives in the molecular genetic analysis of dystrophinopathies: study of a cohort of 471 patients in Argentina.
- Magali Barchuk (Argentina), Familial hyperchylomicronemia syndrome: report of cases in Argentina and importance of the Clinical Biochemistry laboratory for its diagnosis
- Pamela Radic (Argentina), Inactivation of the X chromosome, its importance in the expression of hemophilia in women.

10:00-10:30 break

**Poster session C**, 1<sup>st</sup> floor, Authors of Posters 15 and beyond.

**10:30-11:45. Session VIII, Patients group: Living with a rare disease, examples of resilience.**

- German Franco (Argentina) GM1 Gangliosidosis
- Ana María Forte (Argentina) Family Mediterranean Fever
- Carla Scaramella (Argentina) Hirschsprung Disease
- \*Gastón Varela (Argentina), Phelan-McDermid (PMD) síndrome.

**11:45-13:00 Session IX: Plenary Conferences.**

- Cesar Crespi (Argentina) Education for Rare Diseases in Argentina
- \*Manuel Posadas (Spain) RIBERSER program for Latin America

**13:00-14:15 Lunch Break**

Business lunch (By invitation). Collaboration for rare diseases research. Lecturers room.

**14:15-15:30 Session X: From the FFyB**

Coordinators: Ana Maria Adamo y Liliana Rossetti

- Florencia Giliberto (Argentina), Faculty of Pharmacy and Biochemistry (UBA).  
Molecular Approach to Muscular Dystrophies: Diagnostic Confirmation and its Role in Precision Medicine.
- Cristian Garcia Becerra (Argentina) Development of a novel therapeutic strategy for CoQ10 deficient newborns
- Luis Bredeston (Argentina), Role of the SLC35A3 transporter in congenital glycosylation disorders

**15:30-16:15 Session XI: Selected Oral presentations**

Coordinator: Marcelo Nacucchio

- Enrique Teran (Ecuador), Rare Diseases in Ecuador: A public health perspective.
- \*Domenica Taruscio (Italia), Extended newborn screening for rare diseases: the Italian system.
- Lucía Zavattieri (Argentina), Brucella infection affects functionality of decidual stromal cells and trophoblast.
- Maricel Molina (Argentina), Molecular diagnosis of Thyroid Hormone Resistance Syndrome

16:30-17:00 Closing the 16<sup>th</sup> ICORD annual meeting

---

**Registration:**

On-line: for presentation or on-line attendance. Download "Registration Form" from [www.icord.es](http://www.icord.es) ; [www.cerydh.com.ar](http://www.cerydh.com.ar); and [www.ffyb-uba.org](http://www.ffyb-uba.org)

On-site: Register at secretariat Desk on July 24 (08:00-16:00hs); and July 25 (08:00-13:00hs).

Registration Fee: 50 Euros (Check exchange rate for other currencies)

Registration includes Congress book, entrances to all conferences and poster sessions, simultaneous translation of the conferences, and free coffee services during breaks. Lunch/dinner is not included.

Payment methods: Check the most convenient option.

1. At ICORD webpage: [www.icord.es](http://www.icord.es), credit card only.
2. At FFyB (UBA): [www.ffyb.uba.ar](http://www.ffyb.uba.ar)
3. At CERYDH webpage: [www.cerydh.com.ar](http://www.cerydh.com.ar),

Grants for registration:

-Members of the University of Buenos Aires and CONICeT apply for full registration grants at [www.ffyb.uba.ar](http://www.ffyb.uba.ar)

-Patients, Journalists, (for in person attendance only), apply for full registration grants at [info@cerydh.com.ar](mailto:info@cerydh.com.ar)

-Speakers and posters presenters (one author per poster) receive full grant for registration.

-See grants for registration availability at Sponsors websites. TBC

Companion persons:

-Care givers for patients with special care needs will be admitted free of registration charge.

-Children (<18 years) being affected by rare diseases or companion patients with rare diseases will be admitted free of registration cost and at the sole responsibility of their parents.

---

**Poster presentations: see instructions at websites.**

Free presentations will be admitted in poster format. Please download the form and abstract with instruction at [www.icord.es](http://www.icord.es) ; [www.cerydh.com.ar](http://www.cerydh.com.ar) and [www.ffyb.uba.ar](http://www.ffyb.uba.ar)  
Scientific and Medical information devoted to rare diseases will be admitted upon review from the scientific committee of the congress. Basic and clinical studies, and special patients' cases of rare diseases are encouraged to be exposed at poster session A, B and C.

Poster information from patients' groups activities will be admitted upon review of congress committee. NGOs achievements are encouraged to be exposed at poster session D.

Abstracts will be published together with the Proceeding of the Congress.

---

## **Publications of ICORD 2024 proceedings, TBC.**

All the material received for conferences, poster presentations and satellite symposia will be reviewed and adapted for the publication for the Congress proceedings. Only the version in English language will be admitted. By sending the information to the congress, all the authors agree for its further revision, adaptation, and publication.

ICORD will make its best effort to achieve the editorial approval of the Proceeding of the Congress during 2024.

*Though not all the annual meetings of ICORD have been fully published, please check [www.icord.es](http://www.icord.es) to find similar publications from previous meetings.*

---

### **Information on hotel, transportation and others:**

Registration for the conference does not include accommodation or meals.

The city of Buenos Aires offers a wide range of options and prices that can be accessed through travel agencies or usual electronic means. The Marriot Bonvoy hotel in the "Recoleta" neighborhood is recommended for its proximity to the congress venue.

The congress takes place at Junín 956 of the Autonomous City of Buenos Aires, with several public transportation options. The "Faculty of Medicine" station of line D of the subway is the closest. Taxis or UBER services are cheap and can be requested from the hotel. Consider that traffic in the city is usually slow during rush hours.

Ezeiza airport is 45km from the city center. To go or come to the Ezeiza airport, it is recommended to take a remise or UBER service from the airport, which can be requested from the terminal in the departures sector and paid for with bank means.

The exchange of foreign currency into local currency can be done at the hotel or at authorized banking houses.

The electricity has a current of 220V and the country's electrical system is type I or type C. It is recommended to come with a universal adapter.

---

*-All rights of the material produce by the congress in any format is reserved for ICORD, with exception of the satellite symposia.*

*-Recording or pictures of the sessions is not allowed without permission of the congress organizers.*