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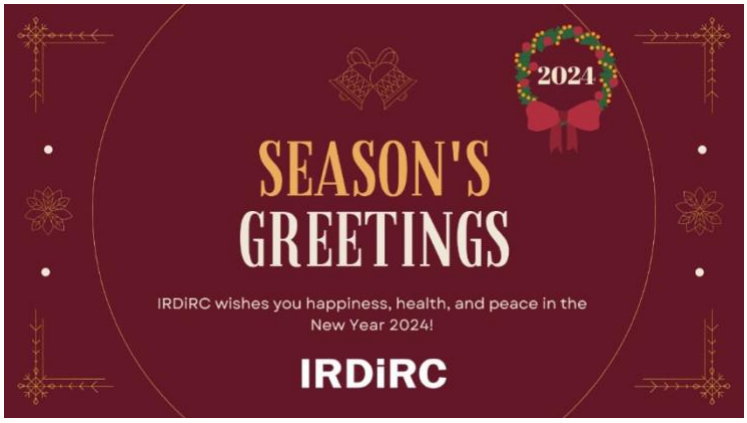
IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

IRDiRC HIGHLIGHTS

Happy New Year 2024!

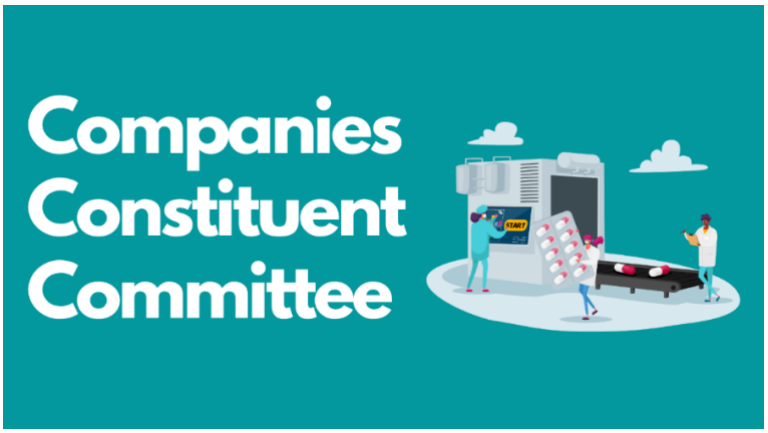


IRDiRC in 2023 – Four new Task Forces, numerous publications, RE(ACT) Congress, World Orphan Drug Congress participation and the list goes on! We couldn't have such a successful year without you! We are sending our sincere gratitude for your fantastic work and involvement towards our common mission for improving the lives of patients with a rare disease!

We wish you a wonderful holiday season and a Happy New Year!

Get to know the Companies Constituent Committee (CCC)!

The **IRDiRC Companies Constituent Committee (CCC)** brings together representatives from 13 pharmaceutical and biotech companies [as of December 2023], to identify common roadblocks to efficient



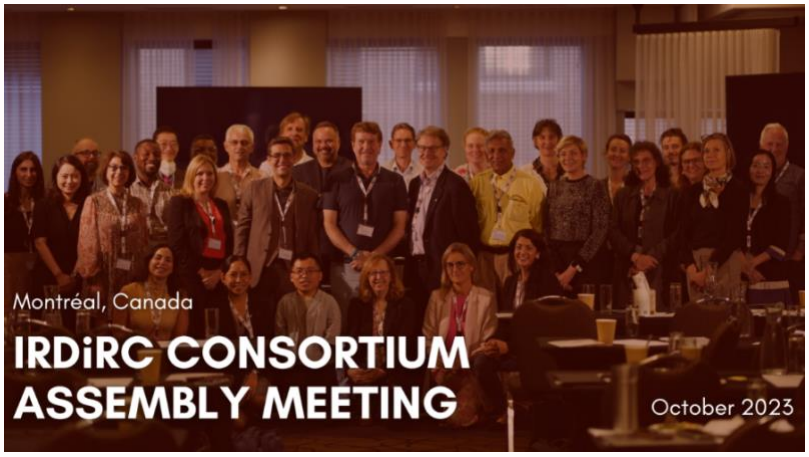
execution of rare disease research, rapid diagnosis, care and pharmacological treatment.
Addressing patient access to diagnostic tools, treatment and care globally is a major focus area for companies. The IRDiRC CCC members stresses the crucial role of natural history of the disease and biomarkers validation in achieving the IRDIRC goals.
Through transparency and sharing of knowledge and skills, the IRDiRC industry partners are serving IRDiRC's mission and goals by bringing together strong expertise, dedication, motivation, and creativity. The CCC group is eager to collaborate with the other IRDiRC committees but also investigates possible external collaborations that would facilitate the implementation or scale up of relevant initiatives with the ambition to forge a better global alignment of scientific and policy recommendations. To foster these collaborations, the CCC has identified a series of key tangible priorities that will give direction to the work over the coming years. The priorities speak to the power of collaboration, the need to develop public private partnerships, to foster global regulatory dialogues and to scale up and implement what works well. The Committee will work on identifying complex problems and prioritizing bottlenecks to address with other stakeholders through dedicated task force and working group.

Private companies have played a major role in **IRDiRC Task Forces** along the years on various themes like **Newborn Screening**, **making rare disease research attractive to other companies**, **targeting shared molecular etiologies to accelerate drug development** or on the likes of the **Orphan Drug Development Guidebook** or **Drug Repurposing**. These dialogues are also incredibly useful to increase understanding between constituents operating under different obligations which is very constructive and conducive to trust.

"We've witnessed many exciting therapeutic and policy progress over the last decade. But the field gets more and more complex. As the awareness for rare diseases grows, more stakeholders are joining the discussion with new perspectives and expectations, meanwhile the need of the patient community remains very high. IRDiRC is a great place to bring up global challenges and work towards solutions" says **Vinciane Pirard**, CCC chair 2023-2026 (SANOFI).

More Information

IRDiRC Consortium Assembly Video

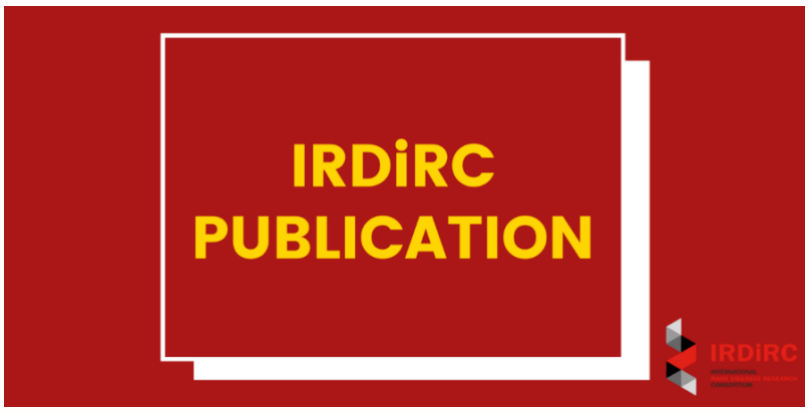


Have a look at the latest video from the **IRDiRC Consortium Assembly Meeting** hosted on 3-4 October in Montréal (Canada) and dive into insightful snippets from different committees discussions!

The members of the Consortium Assembly had a chance to provide live updates on the activities developed within their committees and offered an insight on the priorities for the next months, with the same common goal of driving scientific advancements through research and innovation. Read the **Meeting Report:** here.

Watch the Video

"IRDiRC Drug Repurposing Guidebook: making better use of existing drugs to tackle rare diseases"



IRDiRC proudly presents the new publication developed by the members of the **Drug Repurposing Guidebook Task Force** now available in **Nature Reviews Drug Discovery**. The commentary offers an overview how drug repurposing for rare diseases is possible by organizing the available tools into a standardized framework. The Task Force worked for 18 months with more than 25

experts from the field of RD to help reach the **IRDiRC goal 2**: ***'1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options' by 2027'.***

Read the publication

New Member of the Consortium Assembly

IRDiRC is pleased to welcome **UCB**, represented by **Gabriella Almberg**, Global Head Policy & Public Affairs, Rare Disease Organisation (Denmark), as the new member of the **Companies Constituent Committee (CCC)**.

IRDiRC wishes a very warm welcome to its new member and is looking forward on collaborative future projects.

Change of representation

IRDiRC announces the change in representation for **Global Genes** in the **Patient Advocacy Constituent Committee (PACC). Charlene Son Rigby**, CEO Global Genes (USA) is replacing **Maria Della Rocca**, former Senior Director for Support & Education Programs.

Welcome aboard, Charlene!

IRDiRC EVENTS

Upcoming IRDiRC internal (closed) events:

- **Online Consortium Assembly Meeting**: on **March 6-7, 2024**;
- **In-person Consortium Assembly-Scientific Committees Meeting:** Location - **Shanghai (China)**, on **May 22-23, 2024** with the support of Hope for Rare Foundation, the Chinese Organization for Rare Disorders (CORD), and Fudan University**;**

Collaborative events:

- **2024 Global Rare Diseases Research Symposium & The Second China Rare Diseases Research and Translational Medicine Annual Conference**: Location - **Shanghai, China**, on **May 23-26, 2024**.

IRDiRC representation at events

IRDiRC is delighted to share its strong involvement in this year's edition of the **World Orphan Drug Congress Europe** that took place on 30 October - 02 November in Barcelona (Spain). IRDiRC members and the Scientific Secretariat Coordinator (**Daria Julkowska, Samantha Parker, Anneliene Jonker, Virginie Hivert, Marjon Paasmoij, Dan O'Connor, Violeta Stoyanova-Beninska, PJ Brooks, Clara van Karnebeek, Kelly du Plessis, Mary Wang, Ritu Jain**, **Maria Cavaller Belaubi, Virginie Bros-Facer**, **Durhane Wong-Rieger**) participated in different sessions at the congress highlighting IRDiRC's outputs and mission.

Dr. David Pearce (Sanford Research, USA), IRDiRC chair, offered a presentation on IRDiRC in the International Partnerships and Showcases session. In addition, Dr. Pearce participated in the "Sustainable Global Orphan Markets: Solving the puzzle of rare diseases through international collaboration" session as

panel speaker and was the co-moderator of the session on "Ensuring Patient Access to Cell & Gene Therapies: Operation Warp Speed for Rare Diseases - How to accelerate approval and meet patient needs for C&GT" together with the FCC representative of the US National Institutes of Heath (NIH)/National Center for Advancing Translational Sciences (NCATS), **PJ Brooks**.

NEWS FROM IRDiRC MEMBERS

The Chan Zuckerberg Initiative (CZI): Rare as One

The Chan Zuckerberg Initiative is expanding the **Rare As One Network** and a new cycle will be open next year (including for organisations outside the USA). A call for applications is now in effect.

CZI has funded 2 Cycles so far, and learned how successfully patient organisations are engaging in research collaborations, generating significant impact in their disease areas. Their commitment, to continue to lift up the work of patient communities, is reflected in this new partnership opportunity.

About Cycle 3:

- focus: channelopathies, ciliopathies and inborn errors of metabolism
- open to USA but also other worldwide organisations
- 5-year grant opportunity

Deadline for applications: 22 February, 2024

Apply here

Scientific Call for Proposals AFM-TÉLÉTHON 2024

AFM-Téléthon (France) opens the call for proposals for 2024, open to both French and foreign teams, that aims to support research along the following thematics:

A. Fundamental research and physiopathology of diseases of the neuromuscular system

B. Development of innovative therapeutic approaches for rare genetic diseases

The selected projects will be subject to an agreement with AFM-Téléthon. AFM-Téléthon may decide to finance the selected project under a collaboration which entails a co-ownership of the results.

- Trampoline grant: intended to support young teams or investigators early in their professional career (less than 10 years after thesis, either permanent or non-permanent position), and/or early stage innovative and high risk projects. This grant is awarded for a maximum of 50,000 euros for one year.
- Research project for one year, renewable for a second year, and exceptionally for a third year.
- Post-doctoral fellowship for one year, renewable for a second year.
- PhD fellowship (open to students enrolled in a French university doctoral degree program) for a maximum of three years.

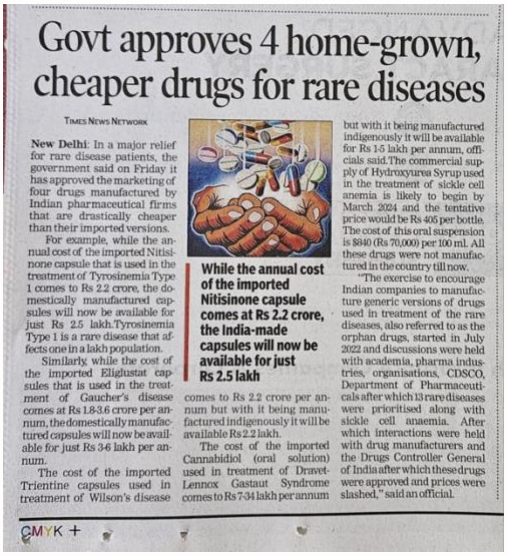
Apply here

Public Meeting on Advancing the Development of Therapeutics Through Rare Disease Patient Community Engagement

On December 14th, 2023, the **FDA** in collaboration with the **Duke-Robert J. Margolis**, MD Center for Health Policy (Duke-Margolis) and supported by a cooperative agreement between FDA and Duke-Margolis, will host a virtual public meeting titled **"Advancing the Development of Therapeutics Through Rare Disease Patient Community Engagement".** The purpose of this public meeting is to highlight and build upon existing actionable approaches for engaging patients, patient groups, and related experts when developing necessary evidence to support rare disease drug approvals.

This public meeting will discuss approaches and opportunities for engaging patients, patient groups, rare disease or condition experts, and experts on small population studies during the drug development process for rare diseases. The meeting will focus on how to best understand patients' experiences living with a rare disease and how to incorporate those experiences and priorities throughout the drug development process.

Registration & More Information



Govt approves 4 home-grown, cheaper drugs for rare diseases

TIMES NEWS NETWORK

New Delhi: In a major relief for rare disease patients, the government said on Friday it has approved the marketing of four drugs manufactured by Indian pharmaceutical firms that are drastically cheaper than their imported versions.

For example, while the annual cost of the imported Nitisinone capsule that is used in the treatment of Tyrosinemia Type 1 comes to Rs 2.2 crore, the domestically manufactured capsules will now be available for just Rs 2.5 lakh.Tyrosinemia Type 1 is a rare disease that affects one in a lakh population.

Similarly, while the cost of the imported Eliglustat capsules that is used in the treatment of Gaucher's disease comes at Rs 1.8-3.6 crore per annum, the domestically manufactured capsules will now be available for just Rs 3-6 lakh per annum.

The cost of the imported Trientine capsules used in treatment of Wilson's disease comes to Rs 2.2 crore per annum but with it being manufactured indigenously it will be available Rs 2.2 lakh.

While the annual cost of the imported Nitisinone capsule comes at Rs 2.2 crore, the India-made capsules will now be available for just Rs 2.5 lakh

The cost of the imported Cannabidiol (oral solution) used in treatment of Dravet-Lennox Gastaut Syndrome comes to Rs 7-34 lakh per annum but with it being manufactured indigenously it will be available for Rs 3-5 lakh per annum, officials said.The commercial supply of Hydroxyurea Syrup used in the treatment of sickle cell anemia is likely to begin by March 2024 and the tentative price would be Rs 405 per bottle. The cost of this oral suspension is $840 (Rs 70,000) per 100 ml. All these drugs were not manufactured in the country till now.

"The exercise to encourage Indian companies to manufacture generic versions of drugs used in treatment of the rare diseases, also referred to as the orphan drugs, started in July 2022 and discussions were held with academia, pharma industries, organisations, CDSCO, Department of Pharmaceuticals after which 13 rare diseases were prioritised along with sickle cell anaemia. After which interactions were held with drug manufacturers and the Drugs Controller General of India after which these drugs were approved and prices were slashed," said an official.

CMYK +

Indian Organization for Rare Diseases (IORD): Government approves 4 home-grown, cheaper drugs for rare diseases

In a groundbreaking move, Indian drug manufacturers have not only successfully developed medicines for Tyrosinemia Type 1, Gaucher's Disease, Wilson's Disease, and Dravet-Lennox Gastaut Syndrome but have also significantly brought down the expenses associated with their treatment.

Expected to be in the market by early 2024, these four medicines are poised to offer even more affordability when compared to their imported counterparts. The monumental cost reduction has been made possible through collaboration between the government and the pharmaceutical industry. The companies have committed to manufacturing these medicines without a profit motive, recognizing the limited demand resulting from the rarity of these conditions. The initiative marks a significant step forward in making rare disease treatments not only accessible but also affordable for the masses.

This is one of the series of efforts undertaken by IORD that was aimed to draw attention to the necessity of producing Make in India orphan drugs, making them more affordable and accessible locally.

More details

Good Off-Label Use Practices (GOLUP) in the European Parliament: Event Report

A meeting was organized in the European Parliament in Brussels on 14 November 2023 by the **European Brain Council (EBC)** to discuss '**Good off-label use practices: Where are we? What next?**'. The meeting was hosted by MEP Stelios Kympouropoulos (EPP, Greece). The meeting touched upon both off-label use and repurposing.

Prof. **Marc Dooms**, Senior Orphan Drug Pharmacist, University Hospitals (Leuven, Belgium) and **Interdisciplinary Scientific Committee Vice Chair**, was adamant to broaden the discussions beyond medicines and include the good-off label use of medical devices, especially for rare diseases. He particularly described the off-label use of devices used for completely different situations: premature babies presenting with patent ductus arteriosus being implanted microvascular plugs or neonates with

congenital heart flow being implanted with a microvascular plug intended for adults. Despite existing guidelines having helped reduce the risk taken with such devices, Prof. Dooms insisted that medical devices being used in completely different situations than those originally intended require the patients' or carer's informed consent. Prof. Dooms acknowledged that off-label use of medicines had yielded good results, some drugs having been repurposed and authorised based on off-label use.

More Information

Frontiers in Medicine: The Changing Focus of Regulatory Frameworks Around the Globe and the Opportunities for Harmonization

IRDiRC is proud to share that **Regulatory Scientific Committee Vice Chair**, **Violeta Stoyanova-Beninska**, is one of the guest editors of the research topic **"The Changing Focus of Regulatory Frameworks Around the Globe and the Opportunities for Harmonization"** in **Frontiers in Medicine**.

As regulation related to the development, registration and monitoring of medicinal products is at different paces in various regions of the world, exchanging of experience and knowledge remains essential to advance the global legislative procedures for the benefit of patients and public health.

The manuscript summary submission deadline is **11 January, 2024**, and the **final manuscript submission is 30 April 2024**.

More Information

Rare Genetic Diseases Research Summit (REDRESS) – 2023

REDRESS is an indigenous and recurrent platform to bring together all rare genetic disease researchers and stakeholders working for the Indian Rare Disease population. This year's second edition took place on November 23-24, in Bengaluru (India) and was organized through a collaborative effort of the the **Tata Institute for Genetics and Society (TIGS)** and **Organization for Rare Diseases India (ORDI)** along with the **The Indian Council of Medical Research (ICMR)**.

More Information

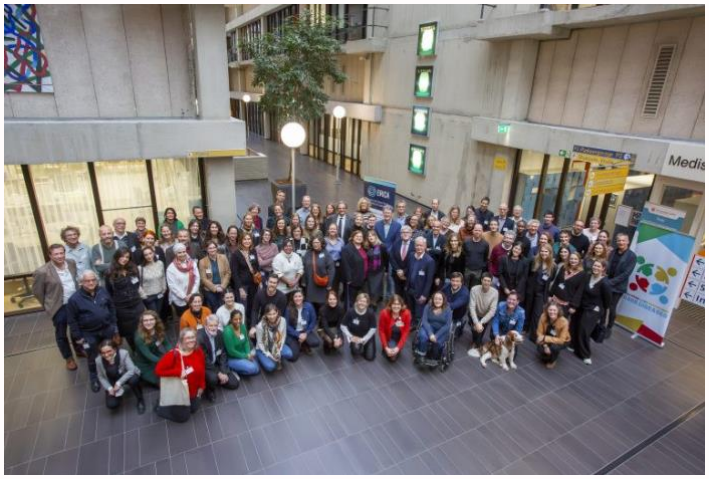
Unmet needs in countries participating in the Undiagnosed Diseases Network International (UDNI): an international survey considering national health care and economic indicators

A new publication focused on **unmet needs** in the countries participating in the **Undiagnosed Diseases Network International**and to which **multiple IRDiRC members (Gareth Baynam, Claudio Carta, Domenica Taruscio, Oleg Kvlividze, Claudia Gonzaga-Jauregui, Saumya Jamuar, Samuel Wiafe)** contributed, is now available in the **Front Public Health Journal.** The publication investigated unmet needs and opportunities for patients with undiagnosed rare diseases. The Developing Nations Working Group of the Undiagnosed Diseases Network International (DNWG-UDNI) conducted a survey, that used a mix of multiple choice and dedicated open questions covering a variety of topics among its members, who were from 20 different nations.

Read the Publication

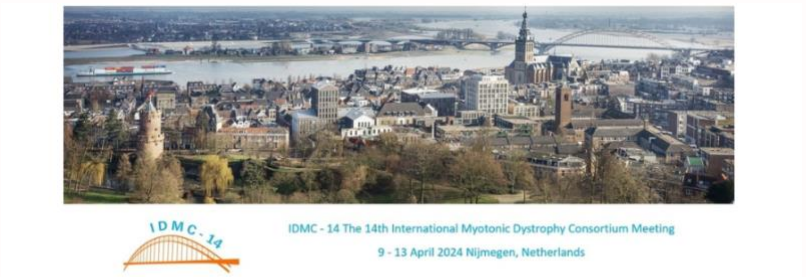
OTHER NEWS

ERICA & EJP RD Joint Conference in Amsterdam



The **European Joint Programme on Rare Diseases** & **ERICA Joint Conference** was held on 21 November 2023 in Amsterdam (The Netherlands). The event aimed to **unite stakeholders in European Reference Networks (ERNs) and Rare Disease (RD) Research**. The meeting brought together medical professionals, researchers, patient advocates, policymakers, and industry representatives who were interested in improving care and advancing research for rare diseases.

More Information



The 2024 edition of the International Myotonic Dystrophy Consortium Meeting (IDMC-14) will be held in Nijmegen from 9-13 April 2024

Nijmegen is the oldest city in the Netherlands and one of the places where the disease-causing repeat expansion was identified in the nineties. **OPTIMISTIC**, the largest myotonic dystrophy trial, was initiated and coordinated in Nijmegen.

The meeting will offer an excellent opportunity to learn, share, and network with all those currently active in the field of myotonic dystrophy: scientists, clinicians, patients, patient representatives, and professionals from the pharmaceutical industry.

IDMC-14 promises to be a truly unique experience, with cutting-edge presentations, interactive sessions, and engaging discussions that will challenge and inspire you. Participants will have the chance to connect with peers and experts from around the world, exchanging knowledge and ideas that will drive innovation and advancement in our field.

Registration & More Information

C-Path: Rare Disease Clinical Outcome Assessment Resource

RD-COAR
Rare Disease Clinical Outcome Assessment Resource

The **Rare Disease Clinical Outcome Assessment (COA)** is an initiative of the **Critical Path Institute's Consortium** with the same name, a public-private partnership that is focused on optimizing COA selection during the drug development for rare diseases. The COAs included in the the Rare Disease COA Resource represent a series of tools that are most commonly used in rare disease research or tools published in the literature, that were available to examine against evidentiary criteria.

What the Resource can do by identifying multiple existing COAs per domain, the considerable time and cost associated with identification of relevant COAs is reduced and made available to all. Evidence from extensive gap analysis on each COA included in this resource can be viewed for each tool individually or

in comparison across several tools in a domain to aid with appropriate COA selection for an invidual research program. The Rare Disease COA Resource can also inform patient advocacy groups of COAs available to measure outcomes of interest in patient registries and natural history studies.

More Information

The Scientific Secretariat of IRDiRC is supported by the European Union through the European Joint Programme on Rare Disease under the European Union's Horizon 2020 research and innovation programme Grant Agreement N°825575.