IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

SAVE THE DATE: IRDiRC Conference and RE(ACT) Congress 2023

IRDiRC is pleased to announce that the next edition of the **5th IRDiRC Conference and 7th RE(ACT) Congress** will be held **in person** in **Berlin, Germany** from **March 15th – 18th, 2023**.

This joint event aims to **bring together scientific leaders, experts, and young scientists from various breakthrough scientific fields to present cutting-edge research, exchange ideas, and discuss policies related to rare diseases research**. Patients and patient organizations committed to research will also be in attendance to share their experiences and perspectives.

The deadline for early registration is October 15th, 2022.

Registration is now open at: https://www.react-congress.org/attendees/registration-2/

On behalf of the Organizing Committee:

- Dr. David Pearce, IRDiRC Consortium Assembly Chair
- Dr. Olivier Menzel, Chairman and Founder of BLACKSWAN Foundation
- Dr. Daria Julkowska, EJP RD Coordinator

More information

New Publication: Recommendations from the IRDiRC Working Group on Goal 3

IRDiRC is proud to note that the **recommendations from the Working Group on Goal 3: Developing methodologies to assess the impact of diagnoses and therapies on rare disease patients** have been published in the *Orphanet Journal of Rare Diseases* in **May 2022**.

The Working Group characterized a set of metrics, tools and needs required for appropriate data collection and establishment of a framework of methodologies to analyze the socio-economic burden of rare diseases on patients, families and health care systems.

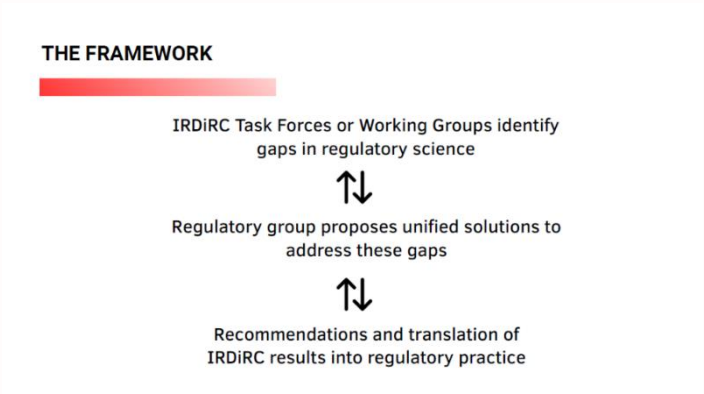
More information

IRDiRC supports 11th European Conference on Rare Diseases & Orphan Products

The **11th European Conference on Rare Diseases and Orphan Products (ECRD) 2022** is being organized by EURORDIS and co-organized by Orphanet (INSERM), with IRDiRC serving as an associate partner. ECRD is a **patient-led rare disease policy event** in which collaborative dialogue, learning and conversation takes place, forming the **groundwork to shape goal-driven rare disease policies**. The **fully online conference** will take place on **June 27th – July 1st** from **14.00 – 18.00 CEST**. **Registration is currently open!**

More Information

IRDiRC Consortium Assembly Validates New Regulatory Scientific Committee



The IRDiRC Consortium Assembly has validated the **establishment of a new Regulatory Scientific Committee**. An open call for member nominations is currently ongoing. The Regulatory Scientific Committee is **part of the overarching IRDiRC goal to accelerate innovative medicines development for rare diseases**. The objectives of the Regulatory Scientific Committee will be to support the **development of IRDiRC activities to address specific regulatory challenges and identify additional topics for regulatory research**.

Health Europa Quarterly publishes interview with Dr. David Pearce

In its **April 2022** issue, **Health Europa Quarterly magazine published an interview with Dr. David Pearce** in an article titled **"Improving care for rare disease patients."** With a **focus on unmet needs in rare disease treatment** and how innovative therapies could offer patients new hope, Dr. Pearce addresses the following questions:

- *What is the role of the IRDiRC within the wider care landscape?*
- *What are the key challenges currently facing rare disease treatment and management?*
- *How has the EU Orphan Regulation introduced in 2000 helped to incentivise research and improve treatment options? What treatment gaps remain?*
- *How could digital or technological solutions help with the diagnosis and treatment of rare diseases?*
- *Are there any notable developments in the world of rare disease management that you think our readers should be more aware of?*

More Information

Leadership and Membership Changes

- Illumina has appointed **Adriana Huertas-Vazquez** (Senior Director, Global Medical Affairs) to replace Volker Liebenberg (Director, Head of Medical Affairs EMEA) on the Companies Constituent Committee (CCC).

IRDiRC IN EVENTS

IRDiRC is holding the following events:

- **Joint IRDiRC Consortium Assembly and Scientific Committees Meeting:** On **June 1st – 2nd, 2022**, an **in-person joint meeting** (with an online attendance option) of the **Consortium Assembly** and **Scientific**

Committees will be held in Paris, France, to discuss IRDiRC and its Committees' strategies and priorities, and provide updates on the activities of the Task Forces and Working Groups.

- **Clinical Research Networks (CRN) Conference:** A joint **Clinical Research Networks (CRN) conference** is planned to be held at the end of **November 2022**. Further details to follow.

IRDiRC was presented at the following events:

- **February 1st**: During the **International Rare Disease Showcase 2022** in a session entitled **"Beyond borders: Empowering an inclusive and international community" by Samantha Parker, IRDiRC Consortium Assembly Vice Chair. Watch the recorded presentation here.**

NEWS FROM IRDiRC MEMBERS

NIH/FDA Workshop on Regulatory Fitness in Rare Disease Clinical Trials

On **May 16th – 17th**, the NIH's National Center for Advancing Translational Sciences (NIH NCATS) and the FDA's Center for Drug Evaluation and Research (FDA CDER) held a **virtual workshop to explore Regulatory Fitness in Rare Disease Clinical Trials,** focusing on academic investigators and those looking to learn how to bridge the gap between academic investigation and the regulatory aspects of drug development.

Workshop recordings (webcasts) are available.

More Information

RDI / EURORDIS Rare Barometer Global Survey on Journey to Diagnosis for Persons Living with a Rare Disease

Rare Diseases International (RDI) is partnering with the EURORDIS Rare Barometer Survey project to launch the **Global Survey on the Journey to Diagnosis for Persons Living with a Rare Disease**. This survey aims to identify factors that influence the process of obtaining a diagnosis and obstacles along the journey. This is a **global survey, available in 26 languages, and is open until June 15th.**

The ambition is to collect relevant responses at the national level and for specific disease areas, while developing an image of the diagnostic journey for our global community.

More Information

Global Genes launches Ukraine Rare Relief Project

In **April**, Global Genes launched the Ukraine Rare Relief Project, to provide **rapid turnaround grants to credible and registered organizations on the ground in the region supporting Ukrainian rare disease families**. Global Genes is also coordinating with partners and providing grants to rare disease organizations who are directly helping displaced rare families with critical and immediate needs in Ukraine and surrounding countries.

More Information

Rare Diseases South Africa (RDSA) introduces Validated Patient Registry

Over the last 12 months, Rare Diseases South Africa (RDSA) has worked on developing an **online (both website and mobile application) patient registry that is confidential and secure** and that is able to collect patient data electronically in a user-friendly way.

At the outset, the registry was intended to allow patients to share their patient journey and learn more about their condition while informing local researchers and clinicians about rare diseases and congenital conditions. However, they have now included an **electronic patient record application** too.

More Information

Funding opportunities with the NIH

On **May 18th**, NINDS, NIH announced a funding call on **Prospective Observational Comparative Effectiveness Research in Clinical Neurosciences**, with several rounds of applications, the first deadline being on **June 17th**.

More Information

Genethon Joins U.S. Bespoke Gene Therapy Consortium

Genethon, a unique non-profit gene therapy R&D organization founded by the French Muscular Dystrophy Association (AFM-Telethon), announced that it has **joined the U.S.-based Bespoke Gene Therapy Consortium (BGTC)** launched in October 2021 by the Foundation for the National Institutes of Health (FNIH) as part of its Accelerating Medicines Partnership® (AMP®) program. The goal is to **speed development of customized (or bespoke) gene therapies for millions of people worldwide suffering from ultra-rare diseases**.

More Information

Indian Organization for Rare Diseases (IORD) undertakes outreach programmes

The Indian Organization for Rare Diseases (IORD) has undertaken **several initiatives recently to enhance the public profile of rare diseases in India**. On **March 24th – 25th**, the Indian Express group (a leading media group in India) organized its **Financial Express Pharma Summit 2022** for which IORD was invited. This included a **session dedicated to rare diseases featuring IORD President and CEO Dr. Ramaiah Muthyala** as the chief speaker. *Recordings of this session are available on the website of the event.*

More Information

OTHER NEWS

MOOC on Diagnosing Rare Diseases: from the Clinic to Research and Back

The **third run** of the **MOOC (Massive Open Online Course) "Diagnosing Rare Diseases: from the Clinic to Research and back"** co-developed by European Joint Programme on Rare Diseases (EJP RD) partners and IRDiRC member **French Foundation for Rare Diseases (*Fondation Maladies Rares*)** opened on **April 18th**. **Registration is free and open at this link**. The topics covered include:

- The diagnostic process and the types of genetic tests available for rare diseases
- The differences in rare genetic diseases patient pathways
- Technological advances for diagnostic research
- The role of collaborative studies and data sharing in rare diseases diagnosis
- The impact of having a diagnosis or lacking a diagnosis on patients' lives
- The role and place of physiopathology approaches as well as social sciences research in the context of rare diseases diagnosis

More information
