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# IRDIRC HIGHLIGHTS

**Happy New Year 2023!**



Our 2023 resolution? To continue with determination and passion working on IRDiRC's mission of advancing research, making a valuable contribution to diagnostics and treatments, and understanding the impact of these, for all people living with a rare disease.

IRDIRC wishes you all the best and a wonderful year!

## **The first International Conference on Clinical Research Networks for Rare Diseases was organized in Paris (France)**

The International Rare Diseases Research Consortium (IRDIRC) and the European Joint Programme on Rare Diseases (EJP RD) jointly organized a conference on **Clinical Research Networks (CRNs) for rare diseases** on **December 1-2, 2022** in **Paris, France**. During this event experts from different continents presented the structure and the activities carried by their respective CRNs. The pathways to stimulate collaboration and interoperability of these networks were discussed and emphasis was put on key topics including:





We are starting 2023 with a new IRDiRC interview, part of the series launched in 2022. This month we introduce you to **Ruty Shai, a member of the IRDiRC Diagnostics Scientific Committee (DSC) and Research Director at Sheba Medical Center (Israel).**



[Watch the video](#)

## Leadership Changes

IRDiRC is pleased to announce the election of **Dr. David Adams (Senior Clinician at the Office of the Clinical Director, National Human Genome Research Institute (NHGRI/NIH), USA)** as the **new Chair** of the **Diagnostics Scientific Committee (DSC)** for a mandate of **3 years**, and the election of **Prof. Clara D.M. van Karnebeek (Professor and Medical Director at Amsterdam UMC, The Netherlands)** as **Vice Chair**.

IRDiRC would like to express its gratitude to the previously appointed Vice Chair, **Birutė Tumiene (Clinical geneticist at the Vilnius University Hospital Santaros Klinikos, Lithuania)** for her contribution and involvement in the work of the Consortium over the past years.

IRDiRC wishes a warm welcome to the new Chair and Vice Chair and it is looking forward to a fruitful collaboration on various rare diseases projects.



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## IRDiRC EVENTS

IRDiRC is organizing the following events:

- **RE(ACT) Congress In-Person:** On **March 15th-18th, 2023**, in **Berlin (Germany)**.

**Registration still open:** <https://www.react-congress.org/attendees/registration-2/>

**Full program:** <https://www.react-congress.org/program/full-program/>

**Submission of abstract poster is extended until January 31st, 2023:**  
<https://www.react-congress.org/attendees/abstract-information/>

IRDiRC internal (closed) events:

- **Joint Consortium Assembly (CA) and Scientific Committees (SC)**  
**Hybrid Meeting:** On **March 14th-15th, 2023** in **Berlin (Germany)**;
- **Consortium Assembly Online Meeting:** on **June 7th-8th, 2023**.



## IRDiRC's previous events



**November 30 - December 01, 2022 (Paris, France)** - A hybrid meeting of the **Consortium Assembly** was held to provide **updates on the activities of the Committees, Task Forces, and Working Groups**, including the planning of IRDiRC priorities and Roadmap 2023.

**January 12, 2023:** An in-person **strategic meeting of the Scientific Secretariat** was held in Paris to discuss and plan the activities of the Consortium and the Scientific Secretariat in 2023 with the Chair and Vice Chair of the Consortium Assembly.

## NEWS FROM IRDiRC MEMBERS

### AFM Téléthon (France) - Call for proposals for 2023

**AFM Téléthon** launched a **scientific call for proposals for 2023**, open both to French and foreign teams. The aim is to support:

- 1. Fundamental Research and Physiopathology of Diseases of the Neuromuscular System**
- 2. Development of Innovative Therapeutic Approaches for Rare Genetic Diseases**

**More information available [here](#) (PDF).**





Along with this call, **AFM Téléthon** also launched a **medical research call in neuromuscular disorders**, having various topics of interest (**patients' management/paramedical care, neonatal screening projects, E-Health and information technologies, assessment of new evaluation criteria, natural history of disease and/or epidemiological studies**). Multiple types of financing are available.

**More information available [here](#) (PDF).**

**Note: Different funding opportunities have different submission timelines. For additional details please check the link below.**

Deadlines and instructions for applications



**Fondation Maladies Rares (France) partners with Alnylam Pharmaceuticals for €20,000 grant award**

**Alnylam 2023 Award - "RNA interferences and rare diseases"**

**Fondation Maladies Rares** is once again partnering with the **Alnylam Pharmaceuticals** to award a €20,000 grant to an innovative research project in the field of rare diseases using RNA interference technology.

The prize will be awarded to an innovative research project in the field of rare diseases and will focus on one of the following themes

- **Translational research developing an RNA interference approach and using an animal or cellular model**
- **Innovative vectoring of RNA interference targeting an organ or a tissue**

The prize will be awarded to a senior researcher, scientific leader of a team attached to a French research organization contributing through his/her research to major advances in the field of rare diseases.

**Application deadline: March 30, 2023, 17h (CET).**

More Information



**AMED (The Japan Agency for Medical Research and Development) launches the call for proposals for 2023**

The main areas of open call are **clinical trial support proposals**, **evidence generation proposals**, and **pathophysiology elucidation proposals**:

- 1. Research on the practical application of innovative drugs for rare and intractable diseases**
- 2. Research field related to the practical application of innovative regenerative and cellular medicine and gene therapy for rare and intractable diseases**
- 3. Evidence Generation Research Directly Linked to Clinical Practice**

#### 4. Research on pathophysiology of rare and intractable diseases

#### 5. Research to Advance the Outcomes of the Undiagnosed Diseases Initiative (IRUD Beyond) Area

Submission deadline: **March 30, 2023**.

[More Information](#)

### **IRDiRC MedTech experts tackle the paucity of Rare Disease Devices**

**MedTech Working Group leader and program lead for personalized medicine at the University of Twente (The Netherlands),**

**Anneliene Jonker**, offered an interview for **Medtech Insight Pharma**

**Intelligence** on the importance of medical devices and technologies used to diagnose, treat or support individuals with rare conditions.

Check out the interview [here](#).



### **Registration for Rare Disease Day at NIH 2023 is now open!**

The **National Institutes of Health (NIH)**'s **National Center for Advancing Translational Sciences (NCATS)** will co-sponsor this year's **Rare Disease Day (RDD) at NIH event** with the **NIH Clinical Center**.

This event aims to raise awareness about rare diseases, the people they affect, and NIH research collaborations that address scientific challenges and advance research for new treatments. RDD at NIH 2023 is a **free event** that is **open to the public**, including **patients, patient advocates, health care providers, researchers, trainees, students, industry representatives and government staff**. This year's event will be in person at the **Natcher Conference Center in Bethesda, Maryland (USA)**. A livestream will be available via **NIH VideoCast** with the event archived for replay afterward.

[Registration and more information](#)

### **Rare Diseases International (RDI) - Universal Health Coverage (UHC) for rare diseases**



With the support of the **UN Office of the High Commissioner for Human Rights**, the **Civil society Engagement Mechanism of UHC2030** and the **NGO Committee for Rare Diseases**, RDI proudly marked the **UHC Day**

(on **December 12th, 2022**) through a key event highlighting the recommendations and the essential next steps towards making **"Health for All"** a reality for the over 300 million Persons Living with a Rare Disease (PLWRD) worldwide.

Check out the recording of the event to learn more about the **main pillars of UHC for rare diseases** and what are the **positive development examples from various regions**: [here](#).

[More Information](#)

### **Instituto de Salud Carlos III (Spain) - More than 100 experts met at the ISCIII to promote advanced therapies in Spain**



The **Spanish Bioindustry Association (AseBio)** and the **Network Biomedical Research Centre (CIBER)** held a conference at the **Chamartín Campus of the Instituto de Salud Carlos III** to **promote the potential of advanced therapies in Spain** and **connect academic research** with the **industrial capabilities of Spanish biotechnology**. The **ISCIII** has stated that the development of advanced therapy medicines is a strategic pillar for the Institute, which has allowed it to make **intensive use of public-private partnership** as a **tool for scientific, industrial and economic transformation**.

**ISCIII representative and member of the IRDiRC Funders Constituent Committee (FCC), Manuel Posada**, presented a lecture about the **history of the rare diseases policies** and the **new call to action** approved by the **European Commission (EC)** at the **XV International Congress of Rare Diseases** which took place in **Murcia (Spain)**, together with **Dr. Eva Bermejo (ISCIII representative)** who held a second lecture about **rare disease diagnosis** and **undiagnosed cases**.

[More information](#)



### **2022 Niemann-Pick Disease Patient-Experts Symposium (China)**

The **Chinese 2022 Niemann-Pick Disease Patient-Experts Symposium** was held on **December 17th, 2022**. The meeting gathered well-known experts in the field to exchange ideas on the latest research results and development trends, interpret cutting-edge progress, and discuss diagnosis and treatment options. The event was hosted by the **China Niemann Pick Care Center** and **Chinese Organization for Rare**

**Disorders (CORD)**.

### **Canadian Institutes of Health Research, Institute of Genetics created Patient Partner Compensation Guidelines**

The **Canadian Institutes of Health Research** developed and implemented **a series of guidelines for patients compensation for participation in research**.

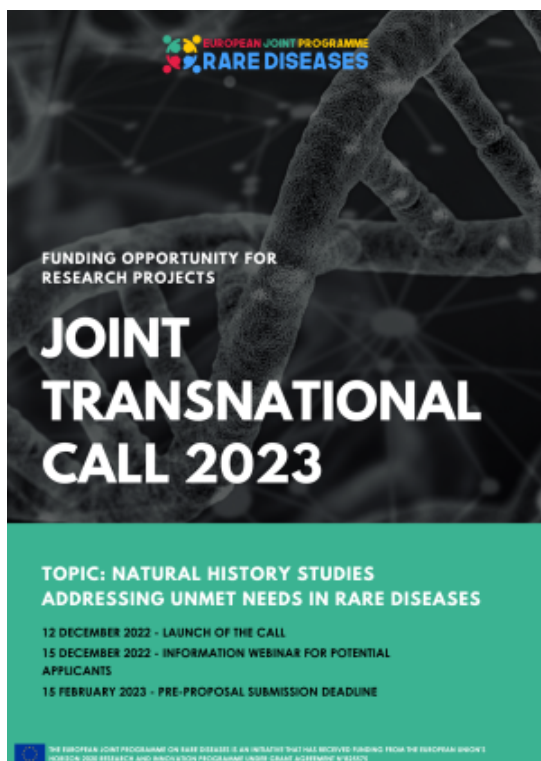


The **Institute of Genomics (IG)** is one of the **13 virtual institutes that make up the Canadian Institutes of Health Research (CIHR)** and its core approach involves starting from **patients** and **civil society**, and working towards **meeting research needs to improve health outcomes**. This approach means that **IG** believes in meaningful inclusion and engagement of patient partners. "**Patient partner**" is an overarching term that includes **individuals with lived experience**, including **patients, caregivers, family, and friends**.

There are many ways for patient partners to engage with **IG**: providing input on specific projects or initiatives, attending meetings or events, speaking publicly, being part of a committee, doing media interviews, writing newsletter articles, and more.

[More information](#)

## OTHER NEWS



### European Joint Programme on Rare Diseases Joint Transnational Call 2023: Now Open!

The **Joint Transnational Call 2023** is a funding opportunity for research projects on the development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases.

This year's topic is: "**Natural History Studies addressing unmet needs in Rare Diseases**".

The aim of the funding opportunity is to enable scientists in different countries to build an **effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise**, with expected impact to **use the results in the future for benefit of patients**.

More information here: <https://www.ejprarediseases.org/jtc2023/>

[More information](#)

### Don't miss the 4th ENMD Congress in Munich (Germany)

The **4th edition of the E-Health & Innovation to Overcome Barriers in Neuromuscular Diseases (ENMD) Congress** will take place on **October 19th-21st, 2023**, in **Munich**, this year's theme is "**Digital biomarkers and digital therapies: New solutions for New Avenues in Neuromuscular Disorders**".





For more information visit:

- [info@fclasssevents.com](mailto:info@fclasssevents.com)
- [www.enmd-neuromuscular.eu](http://www.enmd-neuromuscular.eu)

[More details](#)



SCALING UP PUBLIC-PRIVATE PARTNERSHIPS  
TO ACCELERATE RESEARCH IN RARE DISEASES

## 'Rare Disease Moonshot' – Scaling-up public-private partnerships to accelerate research into world's rarest diseases

A new initiative to **boost research and development** into **rare and paediatric diseases** was launched on **December 8th, 2022**, at the **European Health Summit** in **Bruxelles (Belgium)**.

The **'Rare Disease Moonshot'** is a commitment and collaboration between seven organisations to break down the barriers to finding new treatments and cures for the world's rarest and severe conditions which currently have **no therapeutic options**, and which often affect the youngest patients.

The coalition will work together to **pool expertise, reduce fragmentation in research** and **foster greater collaboration between organisations**. It will bring together an ecosystem of rare disease experts and research to explore opportunities for collaboration and support a range of **public-private partnerships** to:

- Enhance translational research ecosystem to fill the research pipelines with new therapeutic options
- Optimise clinical trials and regulatory pathways for very small patient populations to de-risk and optimise development
- Develop infrastructure to accelerate the journey to diagnosis and treatment

**IRDiRC Scientific Secretariat Coordinator, Daria Julkowska**, was part of the panel **"Building a sustainable R&D ecosystem for rare diseases"** along with other keynote speakers.

[More information](#)

## Orphadata Science granted Global Core Biodata Resource status

On **December 15th, 2022**, **Orphadata Science** was awarded **Global Core Biodata Resource status**, being one of the first batch of resource to be designated by the **Global Biodata Coalition**. **Orphadata Science** (which includes **Orphanet's scientific knowledge base**) was one of the 12 European resources to have been selected following a two-step application process evaluated by a panel of around 50 external reviewers. The next steps imply a discussion with the Coalition's funders and the other designated resources on how to find a sustainable funding model for these resources judged to be keystones in the research data ecosystem.



[More Information](#)

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