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IRDiRC HIGHLIGHTS

Redesigned website and State of Play 2019–2021 now available!

A **newly redesigned, user-friendly** **IRDiRC website** was launched on **June 1st, 2022**, with an **easier navigation panel** to keep up with the latest rare diseases updates.

On **July 5th, 2022**, IRDiRC published the online version of the **2019–2021 edition of the State of Play**, a report



that aims to inform stakeholders at large of **developments in the field of rare diseases research in order to support decisions of policy makers and research funders**. At the same time, the report is focused on informing the rare diseases community at large of the achievements and of observed trends, which shape the future of research and development for rare diseases. It is based on a systematic survey of published articles in scientific journals and press releases.

[More information](#)

IRDiRC at the 2022 Patient Advocacy Certificate Training (PACT)



IRDiRC member **Sanford Research**, in conjunction with the **Professional Patient Advocates in Life Sciences (PPALS)**, hosted the **2022 Patient Advocacy Certificate Training (PACT)** in **Sioux Falls, South Dakota, the United States**, on **May 16th – 19th, 2022**. Dr. David Pearce, Chair of the IRDiRC Consortium Assembly, presented both Sanford Research and gave

an overview of IRDiRC.

[More information](#)

IRDiRC launches the Newborn Screening (NBS) initiative

IRDiRC is launching a discussion on the importance of **Newborn Screening (NBS)** with a focus on covering the topic by including examples of **implementation, usage of results, ethics** and **future state**. As different rules and regulations apply

in different countries, **IRDiRC can bring a major contribution in articulating the status of the science and its applications for all stakeholders**, including patients, physicians, researchers and companies.

Information on this initiative will be updated on the IRDiRC website in due course.



Ongoing calls for member nominations for the ISC and DSC

Calls for new members for the **Interdisciplinary Scientific Committee (ISC)** and **Diagnostics Scientific Committee (DSC)** are ongoing.

- The **Interdisciplinary Scientific Committee** works on cross-cutting aspects of rare disease research, including **data sharing, ontologies, natural history, biobanking, and registries**.

- The **Diagnostics Scientific Committee** brings together **clinicians and experts in genetics, genomics, bioinformatics, molecular diagnostics, and biochemistry** to shorten the diagnostic odyssey often experienced by rare disease patients.

The **mandate** for the Scientific Committees members is **3 years**, with the possibility of **renewal for an additional 3 years**.

[More information](#)

Leadership and Membership Changes

IRDiRC's **Integrating New Technologies for the Diagnosis of Rare Diseases** Task Force announced two new **Co-Chairs**:

- **O'Donnell-Luria, Anne**, Co-Director of the Center for Mendelian Genomics (CMG) at the Broad Institute of MIT and Harvard (USA)
- **Sadikovic, Bekim**, Scientific and Clinical Director, Verspeeten Clinical Genome Centre, London Health Sciences Centre (Canada)

IRDiRC's **Telehealth** Task Force welcomes **new additional members**:

- **Antoniadou, Victoria** – Cyprus Alliance for Rare Disorders, Cyprus
- **Baynam, Gareth** – Rare Care Centre of Western Australia Dept. of Health, Australia
- **Crimi, Marco** – Kaleidos SCS, Italy
- **Dua Puri, Ratna** – Sir Ganga Ram Hospital, India
- **Mueller, Friederike** – Asklepios Hospital, Germany
- **Thong, Meow-Keong** – University of Malaya, Malaysia
- **Tumiene, Biruté** – Vilnius University Hospital Santaros Clinic, Lithuania

IRDiRC IN EVENTS

IRDiRC is holding the following events:

- **Consortium Assembly Online Meeting:** On **September 21st – 22nd, 2022**
- **Consortium Assembly In-Person Meeting:** In **Paris, France** on **November 30th – December 1st, 2022**, back-to-back with the **Clinical Research Networks (CRN) Conference** on **December 1st – 2nd, 2022**
- **Consortium Assembly In-Person Meeting:** In **Berlin, Germany** on **March 14th – 15th, 2023**
- **RE(ACT) Congress In-Person:** In **Berlin, Germany** on **March 15th – 18th, 2023**. Register at <https://www.react-congress.org/attendees/registration-2/>



IRDiRC was presented at the following events:

- During the **ERICA 2nd General Assembly** in **Bologna, Italy** on **20th – 22nd June, 2022**, in a session entitled "**International Rare Disease Research Cooperation from the IRDiRC perspective**" by **Dr. David Pearce**, IRDiRC Consortium Assembly Chair. The presentation can be accessed at the following link: **ERICA Presentation - David Pearce**.

NEWS FROM IRDiRC MEMBERS

**AFM
TÉLÉTHON**
INNOVER POUR GUÉRIR



AFM Téléthon launches the pilot project DEPISMA

The **French Muscular Dystrophy Association (AFM Téléthon)**, in collaboration with the University Hospitals of Strasbourg and the Center Hospitalier Universitaire de Bordeaux, is initiating a **pilot project** called **DEPISMA** for **neonatal genetic screening for spinal muscular atrophy (SMA)** in different regions of France, closely collaborating with the regional health agencies. The start of newborn screening is planned for **September**

2022.

New Grant Funding Opportunity for Rare Disease Research from the FDA

The **Food and Drug Administration's (FDA) Office of Orphan Products Development (OOPD)** is pleased to announce the **availability of funds for fiscal years (FY) 2023 - (FY) 2025 to support clinical trials for rare diseases and conditions**. The purpose of this funding opportunity announcement is to fund clinical trials of products evaluating the **efficacy** and/or **safety** in support of a **new indication** or **change in labeling** to address **unmet needs in rare diseases or conditions**. Additionally, through the funding of collaborative, efficient, and/or innovative clinical trials, FDA expects to increase the number of approved treatments for rare diseases and exert a broad and positive impact on rare disease drug development. Additional information available at **OOPD website**.



[More Information](#)



CIHR Institute of Genetics supports CORD

The **Canadian Institutes for Health Research (CIHR)** supported the **Canadian Organization for Rare Disorders (CORD)** with **virtual and in-person workshops on Canada's strategy for high-cost drugs for rare diseases**.

Additionally, CIHR was also involved in launching different funding opportunities, in partnership with the **Institute for Indigenous Peoples Health** at CIHR on "**Building Capacity for Indigenous First Nations People in Biobanking and Genomics Research**" and a funding opportunity on "**Rare Disease Models and Mechanisms**" linking clinicians with suspected rare disease genes with model organism experts to aid in gene/variant confirmation and to simultaneously provide models to further understand rare disease mechanisms and to test potential therapies.

Western Australian Department of Health made progress on the implementation of the **Rare Care Centre (Centre of Expertise)** at Perth Children's Hospital. The Western Australian Department of Health also undertook the following initiatives:



Government of **Western Australia**
Department of **Health**

- First stage of the new rare diseases knowledge management platform - **UTOPIA - Unlocking Treatment Options, Personalised In-Time Access**, at the Perth Children's Hospital
- Two new **Aboriginal Cadetships for Rare Diseases**, one via the Western Australian Register of Developmental Anomalies (Birth defects and Cerebral Palsy Register) and one via the Rare Care Centre.

[More Information](#)



Meeting Summary of the RDI Informal Side-Event to the 75th World Health Assembly

Rare Diseases International (RDI) would like to thank all who attended the **RDI Informal Side-Event to the**

75th World Health Assembly, which took place in **Geneva, Switzerland** on **May 24th, 2022**.

The event was opened by the **World Health Organization (WHO)** and discussed how **health systems around the world would be strengthened by the development of a Global Network for Rare Diseases**. The WHO also recognised the rare disease community's proposed framework for the Network and affirmed its commitment to addressing rare diseases to achieve Universal Health Coverage.

[More Information](#)

NINDS Interdisciplinary Team Science Grant

The new **NINDS Interdisciplinary Team Science RM1 Funding**

Opportunity Announcement (FOA) encourages interdisciplinary teams of

experts that seek to cross technical and conceptual boundaries through collaboration to achieve ambitious goals for basic, translational, and clinical research questions within the mission of NINDS, the first deadline being on **October 7th, 2022**.



National Institute of
Neurological Disorders
and Stroke

[More Information](#)

FONDAZIONE



Fondazione Telethon (Italy) announces the second round of the "Joint Call for Applications"

Fondazione Telethon (FT) and **Fondazione Cariplo (FC)**, after the success of the first round, continue their alliance to invest in a further joint initiative aiming at **fostering basic research** to support projects focused on aspects of rare diseases that are largely **unknown** or **poorly understood**. Thanks to this Alliance, FC and FT are pleased to announce the second round of the **"Joint Call for Applications"** to solicit

projects focusing on the study of **genes/gene families, proteins, and mRNAs molecules** whose function is unknown in rare diseases, of genetic and non-genetic origin.

[More Information](#)

OTHER NEWS

EJP RD launches Innovation Management Toolbox (IMT)

The **European Joint Programme on Rare Diseases** (EJP RD) is delighted to announce the launch of the **Innovation Management Toolbox (IMT)**, a free-to-use and curated reference **library of resources in rare disease translational medicine** that will provide investigators with self-help resources specific to their needs.



- Learn about the IMT: <https://www.ejprarediseases.org/innovation-management-toolbox/>
- Access the IMT: <https://imt.ejprarediseases.org/>

[More information](#)

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