IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

Results of the Consortium Assembly Leadership Election

IRDiRC is pleased to announce the election of **Dr David Pearce**, President of Innovation, Research & World Clinics, Sanford Health (USA), as the **next Chair of the IRDiRC Consortium Assembly**. Dr Pearce is replacing Dr Lucia Monaco, ex Lead of the Research Impact and Strategic Analysis team at Fondazione Telethon (Italy).

Ms Samantha Parker, Chief Patient Access Officer at InnoSkel (France), is elected **Vice Chair of the IRDiRC Consortium Assembly** and replaces Dr. Pearce in his former role.

*The term of Dr Pearce and Ms Parker will run from **January 1st, 2022** until **December 31st, 2024**.*

More Information

Commentary on 10 years of IRDiRC published in Nature Reviews Drug Discovery

IRDiRC is proud to announce the **publication of a commentary on 10 years of progress and challenges of IRDiRC** in the prestigious journal *Nature Reviews Drug Discovery*. The article **summarizes IRDiRC's vision and goals and highlights its achievements and prospects after its first decade (2011–2021)**. The supplementary information included with the commentary provides a visual summary of IRDiRC's vision and goals, its international reach, the organization of its committees, Working Groups, and Task Forces, an overview of communication activities, and rare disease metrics.

More Information

Call for Members: Task Force on Disregarded Rare Diseases (PLUTO PROJECT)

The Therapies Scientific Committee (TSC) is establishing a **Task Force to characterize specific commonalities amongst a large group of "disregarded" rare diseases**, with the potential secondary aims to identify removable roadblocks that may foster future research and development.

The **PLUTO Project** aims at using an integrated database search approach to:

1. identify and classify the groups of rare diseases that are currently under-represented by academic research and industrial development alike,
2. determine what characteristics they have in common, and – through this analysis –
3. to understand what are the roadblocks that are preventing the chances of seeing effective treatments being developed in the near future.

The TSC is specifically **looking for members to populate the Task Force** with the below expertise in one or more of the following areas:

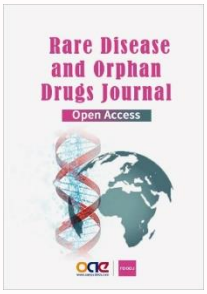
- Drug developers (public and private, for/ no profit)
- Scientists / Clinicians
- Public or private rare disease research funders
- Regulators, Rare Disease Policy
- Patient Advocates

If you are interested in taking part in this activity, please send a CV, biosketch and letter of motivation (one paragraph each) to the **Scientific Secretariat** **before February 11th** with the following reference in the subject line: **TF-IRDIRC-Pluto**.

Only selected candidates will be contacted. Other applications will be kept for potential future use.

More Information

Article on COVID-19 and rare diseases: Reflections and recommendations

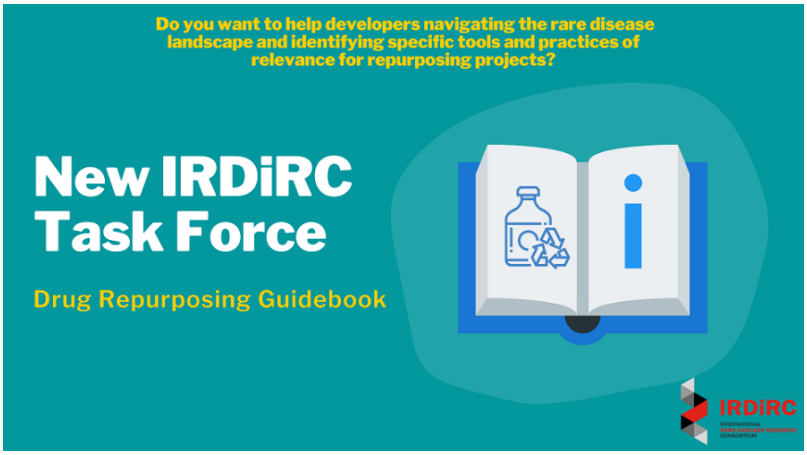


IRDiRC has published the **results of its COVID-19 survey** in the ***Rare Disease and Orphan Drugs Journal***.

The ambitious goals set by IRDiRC by 2027 to fulfill the vision of providing diagnosis and treatments to rare diseases (RDs) patients within one year of coming to medical attention have been challenged by the COVID-19 pandemic. The **article aims to identify the needs and challenges of the RD community during the COVID-19 pandemic** and to understand whether the pandemic would hinder achievement of the IRDiRC goals.

More Information

Call for Members: Task Force on Drug Repurposing Guidebook



The Therapies Scientific Committee (TSC) is establishing a **Task Force on Drug Repurposing Guidebook to help developers (of all kinds) navigating the rare disease landscape and identifying specific tools and practices of relevance for repurposing projects**.

The creation of the Development Guidebook will focus on repurposing approaches, following the same successful methodology used for the Orphan Drug Development Guidebook, i.e. explore incentives, regulatory tools, initiatives, development tools ('building blocks') that exists or are missing for drug repurposing.

The TSC is specifically **looking for members to populate this Task Force** with the below qualities and expertise:

- Drug developers *with expertise in repurposing*
- Scientists / Clinicians
- Regulators
- Public or private rare disease research funders (including investors)
- Patient Advocates
- Patent experts
- Rare Disease Policy experts
- Methodologists (real-world evidence, target validation, AI, in-silico)

Ideally, we would like to see an even participation of experts from different geographical regions.

If you are interested in taking part in this activity, please send a CV, biosketch and letter of motivation (one paragraph each) to the **Scientific Secretariat** **before February 25th** with the following reference in the subject line: **TF-IRDIRC-RepGuidebook**.

Only selected candidates will be contacted. Other applications will be kept for potential future use.

More Information

Leadership and Membership Changes

- **EURORDIS** has appointed **Dr. Virginie Hivert**, Therapeutic Development Director, to replace Dr. Virginie Bros-Facer as their **representative for PACC**.
- **Fondazione Telethon** has appointed **Dr. Stefano Benvenuti,** Public Affairs Manager, to replace Dr. Lucia Monaco as their **representative for FCC.**

*We warmly thank **Dr. Lucia Monaco** and **Dr. Virginie Bros-Facer** for their invaluable contribution to IRDiRC during their tenures.*

IRDiRC IN EVENTS

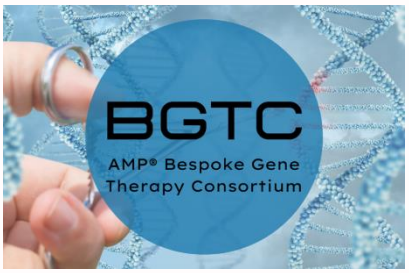
IRDiRC held the following meetings:

- 9-10 December 2021: A hybrid (in-person in Paris and online) meeting of the **Consortium Assembly** was held to announce leadership and membership changes, discuss activities related to the 10-year anniversary of IRDiRC, and provide updates on the activities of the Committees, Task Forces, and Working Groups. Read the report **here**.
- 13 January 2022: 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 2022 with the newly elected Chair and Vice Chair of the Consortium Assembly.

IRDiRC was/is being presented at the following events:

- 14-15 December 2021: During the **APARDO Conference 2021** entitled **"Treating Rare Disease as a Priority"**
- 1-3 February 2022: During the **Rare Diseases Showcase 2022** in a session entitled "**Beyond borders: empowering an inclusive and international community**"

NEWS FROM IRDiRC MEMBERS



Funding opportunities available through Bespoke Gene Therapy Consortium

The Bespoke Gene Therapy Consortium (BGTC), a public-private partnership aimed at **increasing the development of gene therapies for rare diseases,** was launched in **October 2021** by the NIH, FDA, and 15 private organisations. Current funding opportunities include two **request for proposals (RFPs) on human gene therapy**, and the BGTC is also soliciting nominations for rare diseases and disorders to be included in the program.

More Information

InnoSkel awarded Innovation Passport by the UK MHRA

IRDiRC member InnoSkel announced on **December 16th, 2021** that it has been **awarded an Innovation Passport** under the UK Medicines and Healthcare products Regulatory Agency's (MHRA) Innovative Licensing and Access Pathway (ILAP), to pursue **accelerated patient access for its first-in-class treatment for spondyloepiphyseal dysplasia congenita (SEDc)**, a type II collagen disorder.

The **MHRA's Innovation Passport scheme** aims to **accelerate the time to market, facilitating safe, early and financially sustainable patient access to medicines,** through **enhanced input and interactions with the MHRA and other stakeholders**.

More Information

FDA announces 2022 grant funding opportunity for rare disease research

The Food and Drug Administration's (FDA) Office of Orphan Products Development (OOPD) has announced **availability of funds to support natural history studies for rare diseases and conditions**. These studies are intended to provide acceptable data to the FDA that will **substantially contribute to the approval of new products**, or new indications for already marketed products. The next receipt date is **February 15th, 2022**.

More Information

Rare Disease Day at NIH 2022, registration now open!

The National Institutes of Health (NIH) is celebrating **Rare Disease Day at NIH 2022** as a virtual conference on **February 28th**, providing an opportunity to **showcase research on rare diseases by having an exhibit or scientific poster** for thousands of people to visit.

More Information

Fondazione Telethon announces multi-round Call for Research Projects 2021 – 2024

IRDiRC member Fondazione Telethon has announced a Call for research projects that aims at funding **basic and pre-clinical research projects focused on rare genetic diseases** and conducted by researchers working in Italian public or private non-profit research institutions. The call is currently open and the first round closes on **June 30th**.

More Information

OTHER NEWS

UN General Assembly formally adopts Resolution on Persons Living with a Rare Disease and their Families

Following a **sustained campaign by rare disease patient advocacy organisations such as Rare Diseases International** and with the support of several Member States, the United Nations (UN) General Assembly has formally adopted on **December 16th, 2021** with the consensus of all 193 UN Member States the **UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families.** This is the **first ever UN text to give full visibility to the over 300 million persons living with a rare disease worldwide** and calls for action to address the specific challenges faced by these individuals and their families..

More Information

EJP RD Joint Transnational Call 2022 now open

The European Joint Programme on Rare Diseases (EJP RD) has opened its **Joint Transnational Call 2022**, 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. The **pre-proposal submission deadline** is **February 16th, 2022**.

Topic: Development of new analytic tools and pathways to accelerate diagnosis and facilitate diagnostic monitoring of rare diseases

JTC2022 timetable, contact, and more information here: **https://www.ejprarediseases.org/jtc2022/**

More Information