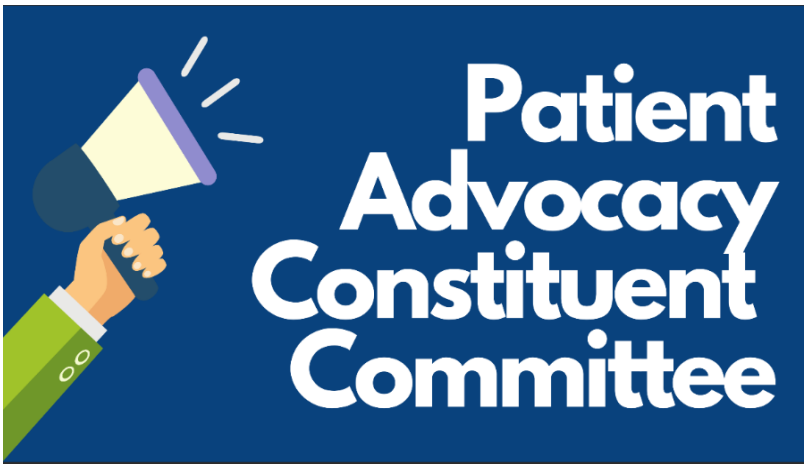
To consult the online version, click here

IRDiRC HIGHLIGHTS

Get to know the Patient Advocacy Constituent Committee (PACC)!



Treatments are often unavailable for rare disease patients, especially in low-and-middle-income countries (LMIC). Reasons for this include lack of financial support for therapies and onerous regulatory requirements for approval of drugs. Other barriers include lack of reimbursement, administrative infrastructure, and knowledge about diagnosis and drug treatment options. The goal of leaving no one behind requires that access to treatments is made available for rare disease patients.

In 2019-2020 the **Rare Disease Treatment Access Working Group** addressed the objective of improving global access to rare disease medicines and published a **list of essential orphan products** for review, recommendations and dissemination.

In 2021-2022 IRDiRC and **Rare Diseases International** **(RDI)** joined efforts to identify the barriers to accessing rare disease drugs in different countries, including high income and LMIC countries. **Cystinosis** and **Cystic Fibrosis** were selected as use cases.

Since 2023, the **Task Force on Impacts and Disease Burden** is developing, operationalizing, and testing a comprehensive framework of holistic, multidimensional, and evolving life-long experiences of patients and families living with a rare disease. This research project builds upon the work of the **IRDiRC Working Group on Goal 3** who produced a **framework of the patient journey** identifying key areas for developing methodologies to assess the impact of diagnoses and therapies on rare disease patients and families.

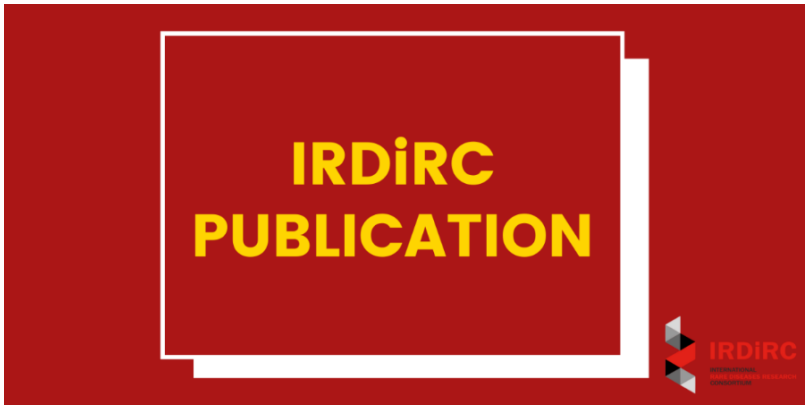
More Information

IRDiRC Interview Series

IRDiRC is delighted to present **Karla Ruiz de Castilla**, **PACC member representative of the Iberoamerican Alliance for Rare Diseases (ALIBER)**. Karla is also the President of **ESPERANTRA**, a non-profit organization founded with the purpose of contributing to the reduction of mortality from cancer, chronic and rare diseases, having a strong advocacy focus on equal access to quality treatment and innovative care. She is based in Peru, South America.

Watch the video

New IRDiRC Publication



IRDiRC is very proud to announce the publishing of the manuscript **"Targeting shared molecular etiologies to accelerate drug development for rare diseases"** summarizing the work of the Task Force on **Shared Molecular Etiologies Underlying Multiple Rare Diseases.**

The article is published in the **EMBO Molecular Medicine Journal** and it is **freely accessible online**.

Link: https://www.embopress.org/doi/full/10.15252/emmm.202217159

More IRDiRC Publications

Membership Changes

Companies Constituent Committee (CCC)

New member

- **Lewis Raynor**, Director Epidemiology, **Biogen** (USA)

Change of representation

- **Vinciane Pirard**, Lead Scientific Advocacy & Insights, Global Medical Affairs, Rare Disease, **Sanofi** (Belgium)

IRDiRC wishes warmest welcome, and is looking forward to a successful collaboration!

Diagnostics Scientific Committee (DSC)

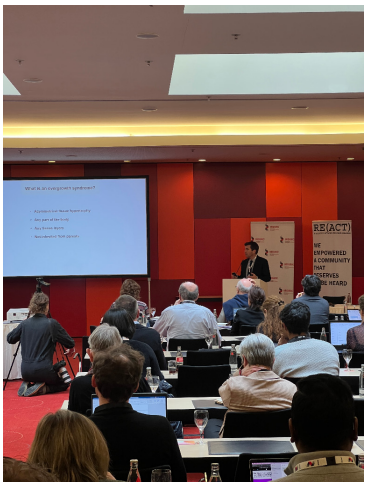
End of mandate

- **Prof. Juergen Reichardt**, **James Cook University**, Australia
- **Prof. Francois van der Westhuizen**, **North-West University**, South Africa

We warmly thank **Prof. Juergen Reichardt** and **Prof. Francois van der Westhuizen** for their valuable contributions and unwavering commitment in the work of the Consortium throughout these past years.

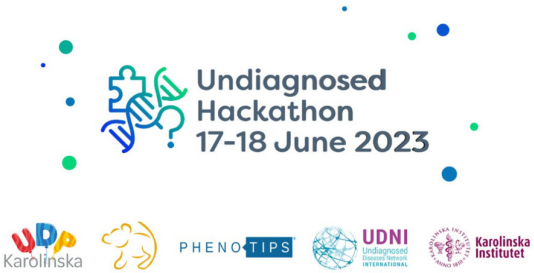
IRDiRC EVENTS

Upcoming IRDiRC internal (closed) events:



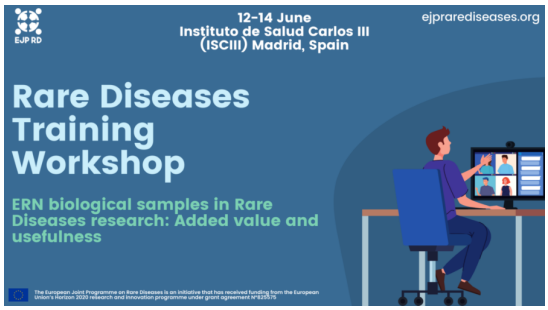
- **In-person Consortium Assembly Meeting:** Location - **Montréal (Canada)**, on **October 3-4, 2023** with the support of **Canadian Institutes of Health Research Institute of Genetics (CIHR-IG)**, **Genome Canada**, **Canadian Organization for Rare Disorders**;
- **Online Consortium Assembly Meeting**: on **December 6-7, 2023 (3-5 PM CET)**;

IRDiRC representation at events



IRDiRC Chair, **Dr. David Pearce (Sanford Health, USA)** attended the **Undiagnosed Hackathon** organized by the **Karolinska Undiagnosed Diseases Program (UDP)**, **PhenoTips**, and the **Wilhelm Foundation** in Stockholm, Sweden, on June 17-18, 2023. The hackathon brought together over 100 global experts in genetics and bioinformatics with a common goal of tackling the complex unsolved cases in new ways, while in the same time raising awareness for undiagnosed diseases.

Dr. Chun-Hung Chan (Sanford Health, USA) gave a virtual presentation entitled **"Challenges and Considerations for Rare Disease Biobanks"** at the **ERN biological samples in Rare Diseases research: Added value and usefulness** training workshop hosted by the **European Joint Programme on Rare Diseases (EJP RD)** in Madrid, Spain, on June 12, 2023.



NEWS FROM IRDiRC MEMBERS

Fondazione Telethon (Italy): Calls for funding opportunities

Fondazione Telethon is pleased to present the following **Funding Opportunities**:

- The third round of the call in alliance with **Fondazione Cariplo**, dedicated to projects focused on aspects of rare diseases that are largely unknown or poorly understood (**Tdark**, https://pharos.nih.gov/), has opened for researchers to apply until **September 27, 2023**;
- The spring 2023 edition of the **"Seed Grants"**, the funding initiative in synergy with Patient's Association, envisages the collaboration with 6 Associations. The consensus meetings happened in the last week of June, with 6 ad hoc panels supporting the evaluation of the received projects.

In addition to the funding opportunities, the foundation is delighted to announce that **AAVantgarde Bio**, a company dedicated to the development of gene therapies for inherited retinal disorders, established in 2021, has announced that it has obtained a 61€ million financing from three major international investment companies: **Atlas Venture**, **Forbion** and **Longwood**. The company is a spin-off of **The Telethon Institute of Genetics and Medicine** (**TIGEM)**, an international research institute based in Naples that is owned and managed by **Fondazione Telethon**. The company's technology builds on existing Adeno-Associated Viral (AAV) vector platforms, one of the main viral vector technologies used in gene therapies applications. The platform is being clinically validated in two inherited retinal diseases: **Usher Syndrome Type 1B associated retinitis pigmentosa (Usher1B)** and **Stargardt disease**. The future pipeline is planned to extend into many disease areas.

More Information

The Foundation for Rare Diseases (France): Call for applications

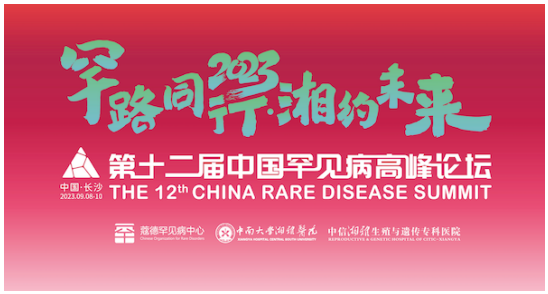
The Foundation For Rare Diseases supports excellent research to understand the causes and pathophysiological mechanisms of rare diseases, to develop new treatments, and to improve the lives of patients. The Foundation has an active scientific policy and launches 4 to 6 calls for projects per year. It offers funding to winning research teams and access to innovative technologies such as new generation sequencing.

Check out the **current calls for projects**:

- Call for **VMOV projects** - **"Research on growth deficits in osteogenesis imperfecta"**, deadline for submission is **July 6, 2023, 5:00 pm CEST**;
- Call for projects **APEHDia** - **"Diaphragmatic hernia research",** deadline for submissions is **September 14, 2023, 5:00 pm CEST**;
- Call for projects **"Identifying therapeutic molecules for rare diseases**, deadline for submissions is **August 31, 2023, 5:00 pm CEST**;
- **"Human and Social Sciences & Sickle Cell Research Grant"** (co-partnership with **Novo Nordisk**), deadline for submissions is **September 19, 2023, 5:00 pm CEST**.

More Information

Chinese Organization for Rare Disorders (CORD): The 12th China Rare Disease Summit on September 8-10, 2023



Inaugurated in 2012, **China Rare Disease Summit** has become the largest and the most comprehensive rare disease conference in China. The annual summit promotes open communications among multiple stakeholders, fosters collaboration, and makes significant contributions to policy advocacy in China.

- The largest and the most comprehensive rare disease conference in China;
- The most robust platform for interactive dialogues among different stakeholders in China;
- A platform for showcasing the latest diagnostic and treatment products in China and abroad;
- A discussion platform for the latest rare disease policies in China and abroad.

For more information please check the website: www.cord.org.cn

More Information

European Union (EU) Research Consortia

The European Union Research Consortia announces new research projects under Horizon Europe.

- **AI-assisted diagnostic and rehabilitative tools for personalised support of children with cerebral palsy -** Unilateral cerebral palsy (UCP) is the most common chronic neurological condition. The EU-funded AInCP project aims to develop AI-assisted clinical decision support tools (DST) capable of providing personalised functional diagnosis, upper-limb (UpL) assessment and home-based interventions for children with UCP. The project will establish a clinical diagnosis and prognosis methodology for personalised UCP treatment using a multimodal approach comprising clinical phenotyping, advanced brain imaging and real-time monitoring of UpL function.

  More information: https://cordis.europa.eu/project/id/101057309
- **EDITSCD – Assessing efficacy and safety of genome EDITing approaches for Sickle Cell Disease** - In the EDISCD project, the consortium will study Sickle Cell Disease (SCD) hematopoietic stem/progenitor cells (HSPC) pathophysiology and improve and benchmark the efficacy and safety of different genome editing approaches for SCD. The disease is one of the most prevalent monogenic diseases in Europe. A single amino acid substitution in the beta-globin chain of the adult hemoglobin (Hb) drives red blood cell sickling and multi-organ damage.

  More information: https://editscd.eu/publications/press-release-editscd/

More Projects

NINDS 2023 Nonprofit Forum

The National Institute of Neurological Disorders and Stroke (NINDS) invites members of nonprofit organizations to an annual **Nonprofit Forum**. The Progress through Partnership Forum provides an opportunity for nonprofit leaders to network with colleagues and to engage in discussions with NINDS staff.

The 2023 NINDS Nonprofit Forum will take place **July 24-25**, virtually and in person at the NIH campus in Bethesda MD (USA). Register today and hold a spot to present a poster on work from your organization (both virtual and in person attendees may share posters).

Registration and more information about the event: https://nindsnonprofitforum.com/

More Information

AMED (Japan): Bringing light to rare diseases with the power of medicine and research

The Japan Agency for Medical Research and Development (AMED)hosted online on July 3, the **Fiscal 2022 Outcomes Conference** titled **"Bringing light to rare diseases with the power of medicine and research"**. As continuous efforts are under way in Japan to address and cure rare and intractable diseases, this conference presented reports on the progress done so far and the outcomes of the latest research and development aiming for novel treatments and diagnoses.

Publication: "Pharmacists are initiators in palliative care for patients with rare diseases"

IRDiRC Interdisciplinary Scientific Committee (ISC) Vice Chair, Marc Dooms, contributed to the article **"Pharmacists are initiators in palliative care for patients with rare diseases"** published in the **Orphanet Journal of Rare Diseases**, highlighting the importance of palliative and terminal care, including the compounding of personalized medication and dispensing of medicinal products and devices in patients with rare diseases.

Read the article

OTHER NEWS

IHI Call Days

The Innovative Health Initiative (IHI) Program Office just pre-published the call for proposals on **"Establishing novel approaches to improve clinical trials for rare and ultra-rare diseases"**.

Check out additional details at the following link:

https://www.ihi.europa.eu/sites/default/files/uploads/Documents/Calls/FutureTopics/DraftTopic_TrialsRareDiseases_June2023.p

df

Don't miss out the recording from the **information webinar**: here.

The **latest drafts** updated on July 12 are available at the following link: https://www.ihi.europa.eu/apply-funding/future-opportunities

More Information

RD Moonshot: Clinical Trials Research Needs Recommendations

The Rare Diseases (RD) Moonshot steering group has just published its recommendations for research funders and health research players from the industry and public sector on research needs that can be best addressed by public-private collaborations to optimise clinical trials in small populations.

The **RD Moonshot** was set up to **boost public private collaborations** in the areas where there are with no treatment options and there is no R&D going on. Modernising clinical trials (design, conduct, regulatory sciences) to make them suitable for very small populations was identified by the RD Moonshot partners as one of the three areas where public private collaborations can add most value.

The **Clinical Trials Research Needs Recommendations** (link: **here**) aim to inform funders of research programmes at national, European and international levels, as well as health research players from public and private sectors who plan projects in the RD white spots areas. The deployment of these recommendations will help prioritising disruptive and impactful actions to close the gap between academic knowledge generation and the next development steps, address the asymmetry of knowledge and skills, and develop regulatory science, amongst others.

More Information

EJP RD News: New interview of Pr. Pierre Emmanuel Gleizes – RIBOEUROPE: The European Ribosomopathy Consortium!

Watch the insightful interview with **Pr. Pierre-Emmanuel Gleizes**, Coordinator of **RIBOEUROPE: The European Ribosomopathy Consortium**.

Funded by the **European Joint Programme on Rare Diseases (EJP RD)**, **Pr. Pierre-Emmanuel Gleizes** sheds light on the remarkable work his team is doing to combat a rare disease and the tremendous potential of European collaboration in achieving their goals.

More information on **RIBOEUROPE here**.

Discover their groundbreaking efforts leading to life-changing results for patients!

Watch the video

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.