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IRDiRC HIGHLIGHTS

Join IRDiRC Diagnostics Scientific Committee!

IRDiRC
Nomination
Call

Diagnostics Scientific Committee (DSC)

DSC is looking for a rare disease diagnostic expert and a rare disease patient/patient advocate who are passionate about improving rare disease diagnosis globally. The ideal candidates should be actively engaged in initiatives, research, or projects related to rare disease diagnostic methods, designing diagnostic systems and processes, and improving existing diagnostic tools or developing new technologies.

Apply before **09 June 2023**

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC's Diagnostic Scientific

Committee (DSC) plays a critical role in

identifying and addressing current and future bottlenecks to **rare disease gene discovery and diagnosis**.

As we work towards improving rare disease diagnosis globally, the **DSC** is currently seeking to expand its membership by welcoming a **rare**

disease diagnostic expert from Africa or Latin America and a rare disease patient/patient advocate from anywhere in the world. We welcome individuals who have a strong interest and involvement in activities or initiatives to improve rare disease diagnosis and are passionate about making a positive impact in the field. We are looking for individuals who can commit to quarterly teleconferences, a yearly face-to-face meeting, and regular committee activities, including email correspondence.

If you meet these criteria, we encourage you to apply and join our global community of experts and individuals who share the same vision and mission. Please note that this is a volunteer membership. To apply, please send us your **resume, bio-sketch, and letter of motivation** to scisec-irdirc@ejprarediseases.org before **09 June 2023**.

IRDiRC Interview Series

IRDiRC is thrilled to present a new video from the **Interview Series with its members**.

This month we have in the spotlight **David Adams, IRDiRC Diagnostics Scientific Committee (DSC) Chair and Deputy Director of Clinical Genomics at NHGRI/NIH (USA).**

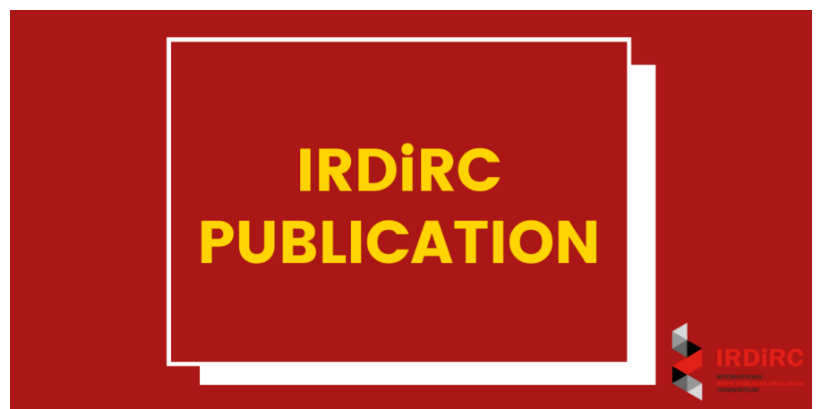
Check out David's answers on what **IRDiRC** means for him and the role it plays for the Rare Disease Community.

[Watch the video](#)

New IRDiRC Publications

IRDiRC announces the publications of two new articles [summarizing the work of the Task Forces](#) on:

- [Clinical Research Networks for Rare Diseases](#)
- [Sustainable Economic Models in Drug Repurposing](#)



The articles are **freely accessible online**.

Towards the International Interoperability of Clinical Research Networks for Rare Diseases: Recommendations from the IRDiRC Task Force.

Published in the [Orphanet Journal of Rare Diseases](#).

Link: <https://doi.org/10.1186/s13023-023-02650-4>

Sustainable Approaches for Drug Repurposing in Rare Diseases: Recommendations from the IRDiRC Task Force.

Published in the [Rare Disease and Orphan Drugs Journal](#).

Link: <http://dx.doi.org/10.20517/rdodj.2023.04>

[More IRDiRC Publications](#)

Membership Changes

Interdisciplinary Scientific Committee (ISC)

Claudia Gonzaga-Jauregui, Assistant Professor at Universidad Nacional Autónoma de México, Mexico

IRDiRC wishes a warm welcome to its new member and is looking forward to a fruitful collaboration on various rare diseases projects and initiatives.

We warmly thank **Dr. Annemieke Aartsma-Rus**, former **Therapies Scientific Committee** member, for her invaluable contribution to IRDiRC during her tenure.

IRDiRC EVENTS

Upcoming IRDiRC internal (closed) events:

- **Online Consortium Assembly Meeting:** on **June 7-8, 2023 (3-5 PM CET)**;
- **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)**;
- **Online Consortium Assembly Meeting:** on **December 6-7, 2023 (3-5 PM CET)**;
- **In-person Consortium Assembly - Scientific Committees Meeting:** Location - **China** (exact city tbc), on **27-28 March, 2024** with the support of **Chinese Organization for Rare Disorders (CORD)**.



IRDiRC representation at events



IRDiRC Chair, **Dr. David Pearce** gave a keynote presentation titled "Advancing rare disease research from discovery to treatment access" at the **19th Orphan Drugs & Rare Diseases Global**

Congress 2023 in London (UK), on 3-4 April, 2023.

Dr. David Pearce also participated in **Hospital Metropolitano's Peds Symposium** where he presented "From the bench to treatment: The complex landscape of rare diseases", held in **San Jose, Costa Rica**.

NEWS FROM IRDiRC MEMBERS

Notice of Upcoming Funding Opportunity: National Pediatric Rare Disease Clinical Trials and Treatment Network

The Canadian Institutes of Health Research – Institute of Genetics is pleased to announce the upcoming **funding opportunity "National Pediatric Rare Disease Clinical Trials and Treatment Network"**.

The **specific objectives** of this funding opportunity are to:

- Develop a platform to support pediatric rare disease clinical trials in Canada;
- Increase the capacity to perform rare disease clinical trials in Canada;
- Attract international clinical trials;
- Foster diversity and inclusion in and improve access to clinical trials;
- Implement processes to acquire data from clinical trials, real world registries, health economics evaluation, and post-marketing surveillance;
- Increase the number of new rare disease drug submissions for authorization of commercialization to Health Canada.



More details on the **grant award** and **application process**: [here](#).

[More Information](#)



AMED (Japan): New Personalized Medical Promotion Program

The Japan Agency for Medical Research and Development (AMED) established a **personalized medical promotion program** as a new application framework. This program includes the following research areas:

- 1) **Early identification research of diseases that are difficult to diagnose or are undiagnosed using short-read whole-genome sequencing;**
- 2) **Innovative biomarker discovery research using multi-omics analysis;**

3) N-of-1+ drug discovery research based on genomic information.

This program corresponds to the basic strategy of Japan's "**Whole Genome Analysis Implementation Plan 2022**", which aims to promote research and drug development, introduce them into daily medical care, and promote new personalized medical care.

[More Information](#)



Orphan Designation: An Overview by EMA

About 26 million people living in the European Union (EU) suffer from a rare disease. **The European Medicines Agency (EMA)** plays a central role in facilitating the development and authorisation of medicines for rare diseases, known also as "orphan medicines".

Don't miss out the video explainer, to which the **IRDiRC Regulatory Scientific Committee (RSC)** Vice Chair, **Violeta Stoyanova-Beninska** also contributed, to learn how **EMA** supports the development of orphan medicines to treat rare

diseases in the EU.

[Watch the Video](#)

The 35th Annual Meeting European Musculo-Skeletal Oncology Society (EMSOS)

The 35th Annual Meeting of EMSOS took place this year on **May 10-12, 2023**, in **Brussels (Belgium)**, and it joined the efforts of all Belgian

University Hospitals and professions represented in the organizing

committee to address this year's theme **"Back to (multidisciplinary) reality: how far do we go?"**

EMSOS
35th ANNUAL MEETING
BRUSSELS | 2023



Marc Dooms, IRDiRC Interdisciplinary Scientific Committee (ISC) Vice Chair offered a speech about **orphan devices** in the session **"Teliagnostics, referral, AI in diagnostics"** on **May 11th**.

[More Information](#)



Rare Diseases Needn't Be Neglected Anymore

India has over 450 types of rare diseases. Lack of awareness and stigma attached to rare diseases have added to the woes of patients suffering from them. The exorbitant cost of therapies makes many patients opt out altogether. Though the Ministry of Health and Family Welfare formulated a National Policy for Treatment of Rare Diseases (NPTRD) in 2017, the

implementation was faced with a lot of challenges. Lack of clarity on the exact number of patients, cost-effectiveness of interventions for rare diseases vis-a-vis other health priorities took a toll on those who suffered from the disease, along with poor awareness among the healthcare staff and lack of proper diagnostic facilities. Check out the interview of **Dr. Ramaiah Muthyala, IRDiRC PACC member representative** and President and CEO of the **Indian Organisation for Rare Diseases**.

[Read the Interview](#)

24th Meeting of the WHO Expert Committee on the selection and use of Essential Medicines

At end of **March 2023**, **Rare Diseases International (RDI)** submitted to **WHO** a report on deliverables of the third year of collaborative partnership:

- (1) consultation and impact on "global description" of rare diseases;**
- (2) in collaboration with IRDiRC, report on status of global access to rare disease medicines;**
- (3) third-year activities and plans related to Global Rare Disease Network of Centres of Expertise.**

RDI is continuing its collaborative partnership with activities on global access to medicines and the global network of CoEs.

RDI participated to the **WHO Expert Committee on the Selection and Use of Essential Medicines Open Session** that



RARE
DISEASES
INTERNATIONAL

was held on **24-28 April 2023 in Geneva (Switzerland)** and **online**, and provided a statement regarding essential medicines for rare diseases.

[More information](#)

OTHER NEWS



THE PUBLIC CONSULTATION ON THE **STRATEGIC RESEARCH & INNOVATION AGENDA** OF THE FUTURE EUROPEAN RARE DISEASES PARTNERSHIP

IS NOW OPEN



This Partnership is proposed for funding under the health cluster of the EU Framework Programme Horizon Europe.

diseases community.

Share your opinion before 9 June 2023.

More information: [here](#).

Now Open: The Public Consultation on the Strategic Research & Innovation Agenda of the future European Rare Diseases Partnership

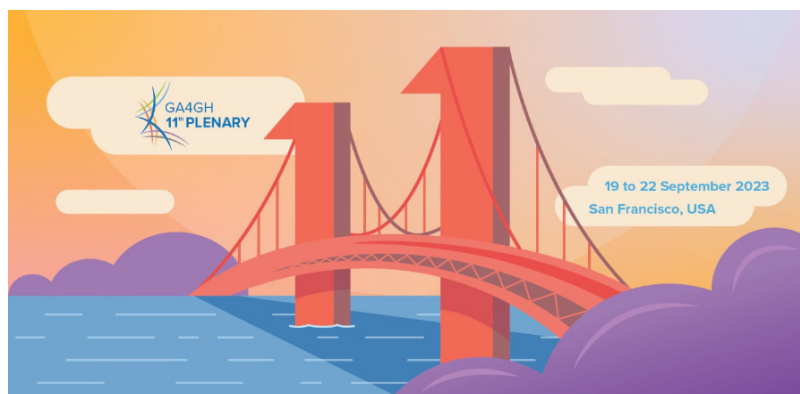
Under **Horizon Europe**, the **European Commission** together with member states and associated countries decided to implement the **Rare Diseases Partnership** which brings a unique vision that aims at leaving no one behind by supporting robust patient need-led research, by utilising and maximising the power of health and research data, by engaging and coordinating regional, national, EU and international alignment in order to accelerate the development of new treatments and diagnostic pathways. To build its strategy on the voice and input of all stakeholders and as part of the preparatory process of this future **Rare Diseases Partnership** (foreseen start in 2024) the **Strategic Research and Innovation Agenda of the RD Partnership** is now open for public consultation. We hope to gather the inputs of all types of stakeholders to make sure that the actions of the partnership will fit the needs and expectations of rare

[Complete the Survey](#)

Registration is now open for GA4GH 11th Plenary

GA4GH 11th Plenary brings together organisations and stakeholders from the genomics and health community for keynotes, talks, and workshops focused on genomic and clinical data sharing issues that pervade diverse industries, disciplines, and communities.

This year the event will take place in **San Francisco, USA**, from **19 to 22 September, 2023**. The first two days focus on internal working discussions between **GA4GH** contributors. On days three and four, the official **11th Plenary** will introduce **GA4GH** to all and highlight the latest advances in responsible global use of genomic data.



Registration open: [here](#).

[More Information](#)



World Duchenne Awareness Day (WDAD) 2023 Theme Announcement

For this year's 10th edition of the **World Duchenne Awareness Day (WDAD)** the theme is "**Duchenne: Breaking Barriers**".

People living with Duchenne and Becker muscular dystrophy (DMD/BMD) face physical, healthcare

and social barriers. This severely limits their ability to participate fully in community life and activities.

On **September 7th**, we call on you to take on responsibility and help break down barriers for people living with DMD/BMD.

On this day, a **WDAD Documentary** will be launched showing the lives of people living with DMD/BMD across the globe and how they are each breaking barriers in their personal lives.

[More information](#)

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.



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