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IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

IRDiRC HIGHLIGHTS

IRDiRC announces Chair and Vice Chair for the Regulatory Scientific Committee

The International Rare Diseases Research Consortium proudly announces the election of **Dr. Anne Pariser (VP, Medical and Regulatory Affairs at Alltrna, USA)** and **Dr. Violeta Stoyanova-Beninska** (**Chair of Committee for Orphan Medicinal Products, EMA, The Netherlands)** as **Chair** and **Vice Chair** of the **IRDiRC Regulatory Scientific Committee (RSC)**.

The **RSC** has been established this **September 2022** to address the regulatory gaps created by the fast moving pace of rare disease innovation and therapeutic development. The RSC includes experts from around the world, representing multiple geographies and stakeholders from regulatory bodies, patient groups, biotech and pharmaceutical industry, public and non-for-profit organizations, clinicians and scientists.

More information

IRDiRC Therapies Scientific Committee (TSC) Nomination Call

The **Therapies Scientific Committee (TSC) would like to broaden the committee membership**, to represent the worldwide rare diseases therapies community in its full width, from early innovation towards regulatory approval and therapeutic support of patients. As such, the TSC has **three openings for new members**, specifically encouraging participants from the following regions: **Africa, Asia, Australasia** or **South America**. The TSC is looking for rare disease experts from different backgrounds with the following qualities:

- Demonstrable track record in contributing to rare disease therapeutic development in either medicines and/or medical devices in Africa, Asia, Australasia or South America (could be **Industry**, **Regulatory**, **Research**, **Health Technology Appraisal**/**Payer**, **Clinical care** or **Patient group**)
- Expertise will be considered from across the therapeutic spectrum, including from prevention, management, quality of life support to curative care
- Must be able to commit fully and actively contribute to monthly teleconferences, a yearly meeting and regular TSC activities, article drafting, and email correspondence

Interested candidates are invited to send their resume, biosketch and letter of motivation to **scisec-irdirc@ejprarediseases.org** before the **December 16th, 2022**. More information about the Therapies Scientific Committee (TSC) at: https://irdirc.org/tsc/

More about IRDiRC Scientific Committees:

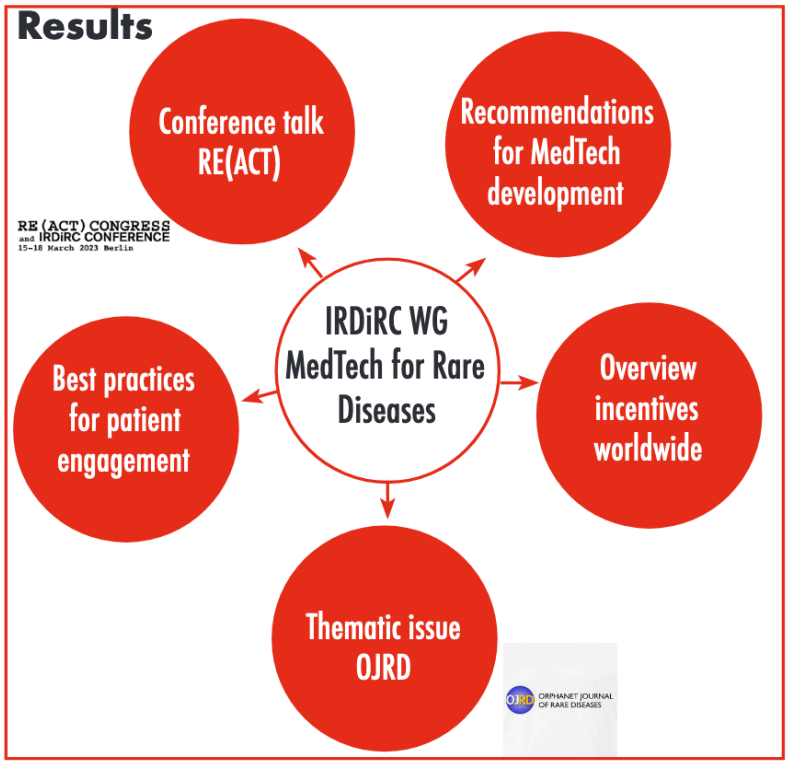
IRDiRC has **four Scientific Committees**, one each for **Diagnostics**, **Therapies**, **Interdisciplinary**, and **Regulatory** aspects of rare diseases research. The TSC is a **multi-stakeholder**, **multi-disciplinary group of experts in medical research and therapy development in rare diseases**. Specifically, the TSC is devoted to support the rare diseases research community in achieving goal 2, "1000 new rare disease treatments by 2027."

More information

Devise – ways forward for medical technology for rare diseases

IRDiRC members and co-leaders, **Marc Dooms** (**Interdisciplinary Scientific Committee**) and **Anneliene Jonker** (**Therapies Scientific Committee**) together with the members of the **Working Group on Medical Technologies for Rare Disease** **(MedTech)** developed a poster focused on ***ways to forward the medical technologies for rare diseases***.

'Medical technologies and devices expected to become increasingly important for rare diseases and rare disease patients. However, in order to further stimulate the development of medical technologies for rare diseases, we need to raise more awareness in both the medical technology and rare diseases community. We also need to gain more understanding of the potential incentives that can stimulate the field.'



See the poster here

Learn more about IRDiRC from its members!

IRDiRC proudly launched **a series of interviews** with its members to discover how the passion for rare diseases research, policy and advocacy drives our members to make an impact globally and serve better the rare diseases community.

This month we would like to introduce you **Marjon Pasmooij**, **Science Programme Manager** at **Medicines Evaluation Board (CBG/MEB), University Medical Center Groningen, The Netherlands,** and **member of the IRDiRC Therapies Scientific Committee (TSC)**.

Watch the video

Leadership changes

IRDiRC is pleased to announce the election of **Gareth Baynam** (**Medical Director at Rare Care Centre, Perth Children's Hospital, Australia**) as the **new Chair** for the **Interdisciplinary Scientific Committee (ISC)** for a mandate of **3 years**.

IRDiRC would like to express its gratitude to the previously appointed chair, **Philip J. (P.J.) Brooks** (**Acting Director Division of Rare Diseases Research Innovation, National Center for Advancing Translational Sciences, NIH, USA**) for his contribution and involvement in the work of the Consortium over the past years.

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

IRDiRC EVENTS

IRDiRC is holding the following events:

Consortium Assembly Hybrid Meeting: On **November 30th-December 1st, 2022** (closed for IRDiRC members), back-to-back with the **International Conference on Clinical Research Networks (CRN)** on **December 1st-2nd, 2022** in **Paris (France)**;

Consortium Assembly-Scientific Committees In-Person Meeting: On **March 14th-15th**, **2023** in **Berlin (Germany)** (closed for IRDiRC members);

RE(ACT) Congress In-Person: On **March 15th-18th, 2023**, in **Berlin (Germany)**; Registration open: https://www.react-congress.org/attendees/registration-2/

Check out RE(ACT) Congress full program

IRDiRC representation at events

RareConversations
Rare Disease Policies: Pioneering the way towards a resilient ecosystem

- **IRDiRC Chair**, **David Pearce**, participated in a panel discussion ***"Partnerships for future: a cooperative approach to address key outstanding challenges"***at the **Rare Conversations Conference** on **October 13th** in **Brussels (Belgium)** and in the **Scientific Advisory Board meeting of the Accelerating Research and Development for Advance Therapies (ARDAT) general assembly** on **October 16th-17th**. **Dr. David Pearce** also presented IRDiRC in a dedicated session at the **11th International Conference on Rare and Undiagnosed Diseases** that took place at the **Medical University of Vienna (Austria)**.

CoRE Scientific Conference
Patients as Partners for Health:
Co-Creating Equitable Access to Health Products and Services

- **Ritu Jain**, **PACC**representative on behalf of **Asia Pacific Alliance for Rare Disease Organisations (APARDO)**, presented IRDiRC at the **DUKE-NUS CoRE Scientific Conference** for the session: ***Building and Using Regional Multi-Stakeholder Platforms to Progress Engagement in the Region.***
- **Samantha Parker, IRDiRC Vice Chair (Innoskel),** together with **IRDiRC CCC representative, Diego Ardigò (Chiesi Farmaceutici SpA)** presented **IRDiRC** at the **Orphan Drug Congress Europe** in **Barcelona (Spain)** in the session: **"How can IRDiRC accelerate rare disease research?".**

NEWS FROM IRDiRC MEMBERS

REMEDi4ALL is building a sustainable European innovation platform to enhance the repurposing of medicines for all

Within the EU-funded project on Drug Repurposing, **REMEDi4ALL**, **a Funders Network** is being created within the next months **comprising different types of funders** who strive to **bring forward Drug Repurposing**. The **initial goal** is to **identify barriers in funding Drug Repurposing**, **propose potential solutions** and **share best practices**.

Funders active or willing to be active in funding Drug Repurposing are invited to join and participate in the debate. The Funders Network is led by **ZonMw**, **the health research funding organisation** in **The Netherlands.**

If you share our passion to enhance the field of Drug Repurposing and want to participate in the debate, please contact **Heleen van der Meer** and **Dunja Huijbers** via **REMEDi4ALL@zonmw.nl** before **December 1st, 2022.**

Read more about the REMEDi4ALL Initiative

The U.S. Food and Drug Administration released a **press release** with **FY 22 funding information** from the **Orphan Products Grants Program** including funding of **11 new clinical trials** and **8 new natural history studies for rare diseases**. FDA also funded **2 contracts** related to **rare neurodegenerative diseases**. Several awards including the 2 contracts and 3 natural history studies support the **Accelerating Access to Critical Therapies for Amyotrophic Lateral Sclerosis Act** (***ACT for ALS***) which recently established the **FDA Rare Neurodegenerative Disease Grant Program** to promote medical product development for rare neurodegenerative diseases such as ALS. Read the **full press release** here.

On **October 17th, 2022**, FDA opened a docket to solicit comments on current funding needs in the **rare neurodegenerative disease space** that could be supported by grants from the **Office of Orphan Products Development**. More information available here. Comments are due by **November 28th, 2022**.

GÉNÉTHON, the AFM-Téléthon laboratory (France), treats its first FKRP patient

A first patient was treated in a **European gene therapy trial** for **limb girdle muscular dystrophy linked to the FKRP gene**. A major step, the result of 30 years of excellent research conducted by **Isabelle Richard**, head of the Progressive Muscular Dystrophies team at the Généthon laboratory.

Led by **Atamyo Therapeutics**, a biotechnology company created by Généthon to accelerate the development of gene therapy for limb girdle muscular dystrophy, the clinical trial is taking place in **Denmark**, **France** and the **United Kingdom**. Its objective is to evaluate the effectiveness of gene therapy associated with an **AAV vector** and the **FKRP gene**.

The first patient was treated by **Prof. John Vissing**, Director of the Copenhagen Neuromuscular Center at the Rigshospitalet National Hospital (Denmark) and principal investigator of this study.

"***The treatment of this first patient is the culmination of years of work for my team and myself. This is a step that we researchers are all waiting for and I hope that this drug candidate will make it possible to stop the disease or reverse its course in patients. From now on, we are continuing, with the same determination, our work to continue to develop gene therapies for other limb girdle muscular dystrophy",*** says **Isabelle Richard**, CNRS Research Director at Généthon.

More Information

The Canadian Institutes of Health Research - Institute of Genetics published a Guidance for Core Elements for Research Consent for Human Genome Research in Canada (in CMAJ)

The **KEY POINTS** highlighted:

- This guidance for policy proposes a core set of elements for documents used to obtain participant consent for human genomics research in Canada.
- The core set of elements comprises the essential components needed to ensure appropriate engagement of patients and other participants in genomics research and in the Canadian Human Genome Library (to be launched in 2023).
- The benefits of a core set of consent elements include rationalization of approval for human genomics research projects by research ethics boards, and increased sharing of genomic and associated health information data across the country.

- Use of a standardized set of consent elements can support the development of the federated Canadian Human Genome Library, in which advanced machine learning methods can be applied to determine which genetic factors contribute to health and disease for those living in Canada.

More information

FDA and NIH Launch Public-Private Partnership for Rare Neurodegenerative Diseases

In September, the **U.S. Food and Drug Administration** and the **National Institutes of Health (NIH)** announced the launch of the **Critical Path for Rare Neurodegenerative Diseases (CP-RND)** – **a public-private partnership** aimed at a**dvancing the understanding of neurodegenerative diseases and fostering the development of treatments for amyotrophic lateral sclerosis (ALS)** and **other rare neurodegenerative diseases**. The FDA and NIH have selected the **Critical Path Institute (C-Path)** as the convener of this partnership.

Read the full Press Release

Global Genes: Know Your Family History Program

Knowing and understanding family history is so important for everyone, particularly communities of color. Last month, **Global Genes**officially launched a new initiative, ***Know Your Family History***, in partnership with the **Black Women's Health Imperative/Rare Disease Diversity Coalition**. Global Genes' goal is to increase the knowledge and understanding of family history and genetic testing, and information and resources are available to help patients on every step of the way. In addition, the Patient Services can assist with specific questions and provide additional information.

More Information

Do not miss Illumina's virtual event "*Shining the light on Rare Genetic Disorders*"

Join Illumina third virtual event, dedicated solely to the **advances of whole-genome sequencing and how it shines the light on rare diseases**. The daily presentations from **leading experts in the field or rare disease** at: **13:00 CET/ 12:00 GMT** from **Monday, 28th November to Wednesday, 30th November, 2022.**

The broad **event agenda** will cover:

- Technical feasibility of implementing whole-genome sequencing into routine clinical care
- Clinical use cases of how whole-genome sequencing advances current standard of care
- Discussions on the need for broadening diversity in genomics data to make genomics relevant to all

...and much more!

Register here

OTHER NEWS

European Joint Programme on Rare Diseases (EJP RD)

The European Joint Programme on Rare Diseases launched a video produced for **ASHG TV** which focuses on the value chain: ***from diagnosis to development of therapies for Rare Diseases***.

Together, EJP RD Members **Dr. Daria Julkowska**, **Dr. Sonja van Weely**, **Dr. Ralph Schuster**, **Anton Ussi**, **Prof. Franz Schaefer**, **Dr. Ana Rath**, **Prof. Rima Nabbout**, **Dr. Roseline Favresse**, and **Prof. Biruté Tumiene** highlight different elements of the EJP RD in the broader context (including genetics and gene therapies), as well as future plans of the programme.

Watch the video

Fondazione Gianni Benzi Onlus - XV Foresight Training Course

Fondazione Gianni Benzi Onlus (Italy) is jointly organising with **Università degli Studi di Bari Aldo Moro, the XV Foresight Training Course *"Boosting research and innovation in a changing regulatory framework"***. It will run as a **hybrid meeting** on **December 15th-16th, 2022** in **Bari, Italy.**

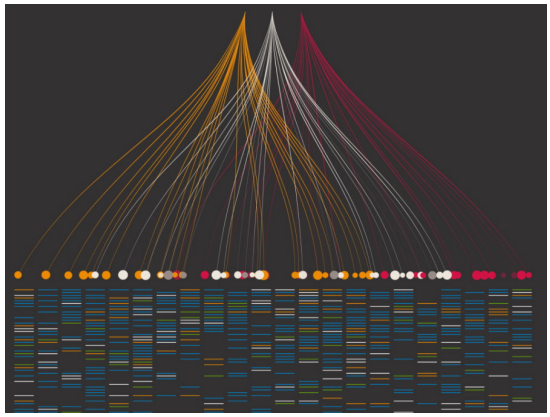
The **course** will be focused on:

- **clinical research**, as expected from the **new Clinical Trial Regulation implementation** and the **Accelerating Clinical Trials in the EU initiative innovative methodologies** including digital and in silico approaches, implemented into all the phases of drugs development, with an insight on the paediatric and rare diseases research
- **challenges and opportunities** of the **use of Real-World Data** and **of a patient-centric model** to boost research and innovation

Relevant stakeholders, including representatives from European Institutions, academia, companies and patients, will provide their point of view.

More Information

Registration open to attend the Third International Summit on Human Genome Editing



The summit is taking place on **March 6th-8th, 2023**at the **Francis Crick Institute**, **London (UK)** and **registration is now open** for **attendance** either **in person** or **online**.

The summit will build on previous events held in **Washington, DC** (2015) and **Hong Kong** (2018), continuing the global dialogue on somatic and germline human genome editing. Major themes for discussion include results from the **first somatic genome editing clinical trials**, **developments in both somatic and heritable genome editing tools** and the **social and ethical considerations** these scientific developments entail.

The three-day event is being organised by the **Royal Society**, **the UK Academy of Medical Sciences**, **the US National Academies of Sciences and Medicine** and **The World Academy of Sciences**. Check out additional details and register below.

More details and registration

European Joint Programme on Rare Diseases Call for Proposals 2023: "Natural History Studies addressing unmet needs in Rare Diseases"

The **EJP RD** has successfully implemented **four Joint Transnational Calls (JTC)** since **2019** to further **help in coordinating the research efforts of European**, **associated and non-European countries in the field of rare diseases** and **implement the objectives of the International Rare Disease Research Consortium**. These actions are following the ten Joint Transnational Calls for rare diseases research projects launched previously by the ERA-Net E-Rare since 2006.
A number of **national** and **regional funding organisations** will participate in the **EJP RD Joint Transnational Call 2023** and **will fund multilateral research projects on rare diseases**. Final decision on participating funding agencies will be issued before the launch of the call. The call is expected to be opened simultaneously by multiple funding organisations in various countries/regions.

More information

ESPERANTRA: "*Clinical Research, hope for patients*".

We are honoured to share this special report made by **Latina Televisión (Peru)** together with **ESPERANTRA (Peru)** which talks about the need for more clinical research on patients with cancer and rare diseases through the testimony of Aida, a patient suffering from a rare form of nasal cancer.

Watch the video

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