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IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

Happy Rare Disease Day from IRDiRC!

What is new?

IRDiRC HIGHLIGHTS

21st Meeting of the IRDiRC Consortium Assembly

21-22 JANUARY 2021

The IRDiRC Consortium Assembly met on 21-22

January 2021 to review the 2021 roadmap

activity proposals. Following the activity prioritization vote, we are glad to announce that IRDiRC will implement two new activities:

- **Big Bang Project – Drug Repurposing Guidebook**

The objective of the Drug Repurposing Guidebook is 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 (https://www.nature.com/articles/d41573-020-00060-w), i.e. explore incentives, regulatory tools, initiatives, development tools ('building blocks') that exists or are missing for drug repurposing.

- **PLUTO PROJECT - Disregarded Rare Diseases**

The PLUTO project aims at using an integrated database search approach to (1) identify the rare diseases that are not currently receiving attention by academic research and industrial development alike, (2) find what characteristics they have in common, and -through this analysis- (3) to understand what are the roadblocks shrinking the chances of seeing effective treatments developed for these diseases in the near future. Based on the results of this analysis, the working group also aims at providing potential recommendations to funders and developers to overcome existing limitations and roadblocks for research and development for these "disregarded" diseases.

These two new activities will complement the Task Forces on «Integrating New Technologies for the Diagnosis of Rare Disease» and «Shared Molecular Etiologies» (see below) to form the final IRDiRC 2021 Roadmap of activities.

Task Forces - now starting!

We are glad to announce the opening of two new task forces.

- **Integrating New Technologies for the Diagnosis of Rare Disease**

  Aiming to identify the most clinically beneficial combination(s) of metabolomic and genomic tests coupled with artificial intelligence methodologies, which would then be prioritized for development of diagnostic standards.

- **Shared Molecular Etiologies**

  Aiming to address and document the existing challenges in adapting the basket trial approach used in molecularly targeted oncology clinical trials to drugs targeting shared molecular etiologies underlying multiple rare diseases.

If you are interested in becoming a Task Force member, the call for candidates will remain open until March 1st, 2021. Please send your CV to the Scientific Secretariat of IRDiRC at scisec-irdirc(a)ejprarediseases.org

The first meetings are expected to start in April 2021. Click below for more information.

More Information

IRDiRC Conference & RE(ACT) Congress

13-15 JANUARY 2021

The IRDiRC Conference & RE(ACT) Congress gathered over **230+ attendees** during the two days. The sessions were inspired by the IRDiRC activities and task forces and engaged the audience in lively

panel discussions with patients' representatives and the session the speakers.

Recordings of the conference are available for six months **here**.

Session A: "Presentation of the Galaxy Guide & Hands-on"

Session B: "Rare Diseases Foresight: Panel discussion (EU/America/Asia/Australia)"

Session C: "Diagnostic, WGS, artificial intelligence, new technologies"

Session D: "Molecular etiology of RD, innovative clinical trials, precision medicine"

Session E: "Advanced therapies: gene editing, cell therapy"

Session F: "Patients as drivers in drug development and clinical trials"

Session G: "Access to diagnostic and drugs for all"

Session H: "Methodologies to assess the effect of diagnosis and therapies on RD patients"

Leadership and Membership Changes

- **Dr. Daniel O'Connor**, MHRA, UK, was elected new Chair of the **Therapies Scientific Committee** for a 3-years mandate. Congratulations Dr. O'Connor!
- We would like to thank **Dr. Diego Ardigò** and **Dr. Virginie Hivert** for serving as Chair and Vice Chair of the **Therapies Scientific Committee**. Their commitment was essential to the work developed by the TSC in the last six years.
- **Dr. Diego Ardigò** is replacing **Dr. Andrea Chiesi** as representative of Chiesi in the **Companies Consitituent Committee**.
- We are happy to welcome **Congenica** as new member of the **Companies Consitituent Committee** and the **Consortium Assembly**. Congenica will be represented by **Dr. Christina Waters**.

Diagnostics Scientific Committee Nominations

We warmly welcome the new members of the Diagnostics Scientific Committee and wish them a fruitful collaboration:

- **Professor Alain Verloes**, Hôpital Robert Debré, Lyon, France
- **Professor Guillem Pintos-Morell**, Vall d'Hebron Research Insititute, Barcelona, Spain
- **Dr. Birute Tumiene**, Vilnius University Hospital, Vilnus, Lithuania
- **Dr. Helen Malherbe**, Rare Diseases South Africa, Cape Town, South Africa

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IRDiRC IN EVENTS

IRDiRC held/will hold the following meetings:

- 15 March 2021: **22nd IRDiRC Consortium Assembly meeting**
- 21-22 January 2021: **21st IRDiRC Consortium Assembly meeting**

To see the available reports and to get more information press the button below.

More Information

IRDiRC has been/will be presented in the following events:

- 12-13 January 2021: **European Joint Programme on Rare Diseases Policy Meeting**
- 18 February 2021: **Chiesi Rare Disease Day Event**
- 12 March 2021: **Congress of the Italian Society of Pharmacology**

OTHER NEWS

Heritable Human Genome Editing Rare Disease Week Discussion

Heritable human genome editing has been proposed as a means of helping parents avoid passing genetic diseases to future generations. But can heritable human genome editing be used safely?

On **February 26, 2021** at 9:00 am EST (2 pm GMT / 3pm CET / 10 pm CST), join members of the International Commission on the Clinical Use of Human Germline Genome Editing for a 90 minute online discussion of the implications of this technology for genetic disease and disability communities.

This event is free and registration is not required.

More Information
