IRDiRC Newsletter 9. September - October 2019

RE(ACT) congress - IRDiRC conference 2020

EARLY REGISTRATION IS OPEN, SUBMIT YOUR ABSTRACT!
List of speakers is available here.
11 - 14 March 2020, Berlin, Germany

The BLACKSWAN Foundation and IRDiRC, the International Rare Diseases Research Consortium, are glad to announce the RE(ACT) Congress and IRDiRC Conference 2020. We look forward to welcome scientists, researchers, patient organizations, and patients in Berlin.

IRDiRC’s State of Play of research report

This IRDiRC’s report is a compilation of information published in scientific journals and press releases over the period of September 2015 to June 2018. The report aims to inform stakeholders at large of developments in the field of rare diseases and support decisions of policy makers and research funders, as well as to inform the rare diseases community of the achievements and observed

New IRDiRC Recognized Resource: The Model Organism Aggregated Resources for Rare Variant ExpLoration (MARRVEL)

MARRVEL is a search engine that collects data from a number of human genomics and model organism genetics databases and displays it in a rapid user-friendly format. It was created in 2017 by a group of researchers in the Undiagnosed
Read more trends which shape the future of research and development for rare diseases.

Read more Disease Network at Baylor College of Medicine as a one-stop data collection tool to facilitate the use of public genetic resources to prioritize rare human gene variants for study in model organisms. MARRVEL is available online and is continuously updated.

IRDiRC New Publications

IRDiRC has three new publications:

- The use or generation of biomedical data and existing medicines to discover and establish new treatments for patients with rare diseases - recommendations of the IRDiRC Data Mining and Repurposing Task Force. Noel T. Southall et al., 2019 - Task Force Data Mining and Repurposing
- Model consent clauses for rare disease research. Minh T. Nguyen et al., 2019 - Task Force Model Consent Clauses for Rare Disease Research
- Ethical, legal, and social issues (ELSI) in rare diseases: a landscape analysis from funders. Adam L. Hartman et al., 2019. Working Group on ELSI.

Conference Presentations on IRDiRC

Here are the past and upcoming conferences and meetings on IRDiRC:

- September 20-22, 2019 - The 8th China Rare Disease Summit - Shenzhen, China
- October 19-20, 2019 - 2019 China Conference on Rare Diseases (CCRD2019) - Beijing, China
- October 19-21, 2019 - APARDO Summit 2019 Regional Collaboration for Global Change – Taipei, Taiwan
- October 28-30, 2019 - XX Convention Scientifica, Fondazione Telethon. Riva del Garda, Italy
- November 11-13, 2019 - The XIV meeting of the International Conferences on Rare Diseases and Orphan Drugs (ICORD) - Tel Aviv, Israel
- November 12-14, 2019 - World Orphan Drug Congress 2019 - Barcelona, Spain
- November 21 - 22, 2019 - IRDiRC Consortium Assembly, Face-to-Face Meeting - Paris, France

IRDiRC new members

IRDiRC is pleased to announce three new partners in the Consortium:

- The French National Institute of Health and Medical Research (Inserm) - the leading European organization for health research, and the coordinating institution of the European Joint Programme on Rare Diseases (EJP RD). Dr Catherine Nguyen, the Director of research of the Thematic Institute Multi-Organisms “Genetics, Genomics and Bioinformatics” at INSERM, was nominated as representative to the IRDiRC Consortium Assembly and Funder Constituent Committee.
- YposKesi - the first French industrial pharmaceutical company dedicated to producing gene and cell therapy drugs for rare diseases. Prof Jean-Pierre Gaspard, Executive Chairman at YposKesi, was nominated as
representative to the IRDiRC Consortium Assembly and Company Constituent Committee.

- **Eloxx Pharmaceuticals, Inc.** - a clinical-stage biopharmaceutical company developing novel RNA-modulating drug candidates designed to treat rare and ultra-rare stop codon diseases. Dr David Snow, Chief Business Officer at **Eloxx Pharmaceuticals Inc.**, was nominated as representative to the IRDiRC Consortium Assembly and Company Constituent Committee.

**IRDiRC video**

Dr. Annemieke Aartsma-Rus’s video on IRDiRC

Dr. Aartsma-Rus is a professor in translational genetic at the Leids Universitair Medisch Centrum and a member of the IRDiRC Therapies Scientific Committee. In this video she tells us why IRDiRC is needed in the rare disease field and why it is important to have different stakeholders collaborating.