http://www.irdirc.org/

IRDiRC Newsletter 13. December 2019

Join a major event on rare diseases in 2020:
The RE(ACT) Congress and IRDiRC Conference
11-14 March 2020, Berlin – Germany.
Early bird registration closes on 20 Dec 2019!

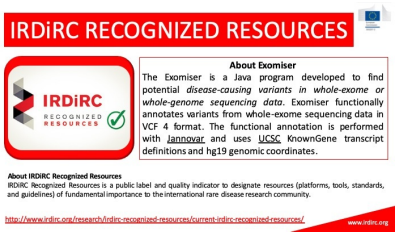
Young and professional scientists, patients & patient representatives, and other rare diseases stakeholders are welcome to discuss latest cutting-edge research, innovation, and policy with the rare disease leaders.

The scientific program – inspired by IRDiRC principles, achievements, and activities foreseen in 2020- encompasses topics like diagnosis & artificial intelligence, molecular etiology, innovative clinical trials, patient engagement and drugs for all. Register and submit your abstract, there is still time!

Blackswan Foundation and IRDiRC in collaboration with EJP RD organize this exceptional joint edition.

Join our social media campaigns by copy and paste the texts here.

IRDiRC highlights



Privacy-Preserving Linkage of Genomic and Clinical Data Sets. Dixie B. Baker, Bartha M. Knoppers , Mark Phillips , David van Enckevort , Petra Kaufmann, Hanns Lochmuller, and Domenica Taruscio. *IEEE/ACM Transactions on Computational Biology and Bioinformatics, 2019.*

Task Force: ***Privacy-preserving data linkage.***

The **Exomiser** is a Java program developed to find potential disease-causing variants in whole-exome or whole-genome sequencing data. Exomiser functionally annotates variants from whole-exome sequencing data in VCF 4 format. The functional annotation is performed with Jannovar and uses UCSC KnownGene transcript definitions and hg19 genomic coordinates. The Exomiser was developed by the Computational Biology and Bioinformatics group at the Institute for Medical Genetics and Human Genetics of the Charité - Universitätsmedizin Berlin, the Mouse Informatics Group at the Sanger Institute and other members of the Monarch initiative.

IRDiRC meetings

The **IRDiRC Consortium Assembly (CA)** gathered for its fall face-to-face meeting on Nov 21 - 22 at the Inserm Biopark in Paris, France. The purpose of this meeting was to define the workplan for the new year with the formal approval of the Roadmap 2020, the introduction of new IRDiRC members, and share updates on the activities carried out by our members' organizations this

past year (2019) in the different countries. There were two parallel sessions (Funders with Companies; Patient Advocates) and a plenary session where we had the opportunity to define common strategies to improve our engagement with patient organizations, further increase our working network as consortium toward new stakeholders, and increase our visibility by sharing our vision across different communities.

Work Plan for 2020

Several new IRDiRC activities, prioritized by the Consortium Assembly, will be conducted by dedicated task forces or working groups in the upcoming year.

IRDiRC actions:

- **Shared Molecular Etiology.** Grouping rare disease patients based on the underlying molecular etiology has the potential to in effect reduce the numbers of disease, while greatly increasing the number of patients gaining access to clinical trials. The focus of this activity is to adapt the molecular targeted basket trials approach from oncology and apply it to drugs that target shared molecular etiologies underlying multiple rare diseases.
- **Rare Diseases Treatment Access.** Treatments are often unavailable for rare disease patients, especially in low-and-middle-income countries and that for a variety of reasons. The goal of *leaving no one behind* requires that access to treatments be available for rare disease patients throughout the world. To deliver this goal, the working group will address barriers to accessing RD drugs in various countries and compile a list of standard-of-care products for RDs to make it available for all countries.
- **Chrysalis Project.** The goal of this activity is to identify: (1) key criteria that would make RD research more attractive to industry; (2) gaps/opportunities in the current funding landscape to delivery against the criteria; (3) other non-financial barriers related to attractiveness of meeting such criteria. Ultimately, this will provide opportunities for expanding the number of treatable RDs through identification of diseases that already respond to most criteria used to inform industry R&D decisions for orphan drugs.
- I**ntegrating New Technologies for the Diagnosis of RD.** Identify the most clinically beneficial combination(s) of metabolomic and genomic tests coupled with artificial intelligence methodologies, which would then be prioritized for the development of diagnostic standards. This will ultimately lead to the more rapid adoption of new techniques/technologies and increase the benefit to patients above the current diagnostic algorithms.

Call for experts will be published on the IRDiRC website in early 2020

Stay tuned for our newsletter editions in 2020,
since we will be developing specific focus on each of these activities!

IRDiRC new members

IRDiRC is pleased to announce three new members in the Consortium as part of the **Therapies Scientific Committee (TSC), welcome!**

- *Anneliene Jonker* is the Funding Officer of the TechMed Centre, at the University of Twentes, The Netherlands. In this position, she is responsible for setting up funding strategies for different researchers in the personalized medicine-, medtech, and rare diseases domain.
- *Janet Maynard* is the Director of the Office of Orphan Products Development (OOPD) at the FDA, USA. In her role, she oversees legislatively mandated designation and grant programs intended to promote the development of products for rare diseases. In addition, she leads the coordination of the cross-cutting rare disease issues and the engagement with patients, sponsors, and other stakeholders.
- *Daniel O'Connor* works in the licensing division of the MHRA, UK. He is the UK representative on the Committee for Orphan Medicinal Products (COMP) and observer on the EMA's Oncology Working Party. He has also contributed as a COMP to member to the EMA's Scientific Advice Working Party (SAWP) and the Patient and Consumer Working Party (PCWP). He is

on the editorial board of the journal Expert Opinion Orphan Drugs.

IRDiRC members spotlights

Dr. Daria Julkowska won the EURORDIS Black Pearl 2020 - European Rare Disease Leadership Award

IRDiRC congratulates Dr. Daria Julkowska for having been awarded of the EURORDIS Black Pearl - European Rare Disease Leadership Award! **Congratulation for this important achievement.**

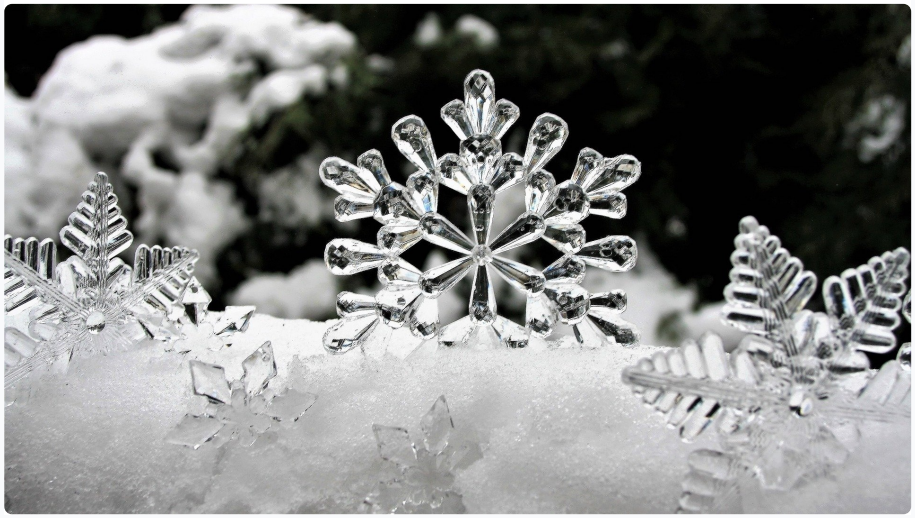
Prof. Annemieke Aartsma-Rus won the EURORDIS Black Pearl 2020 - Scientific Award

IRDiRC congratulates Prof. Annemieke Aartsma-Rus for having been awarded of the EURORDIS Black Pearl - Scientific Awards 2020! **Congratulation for this important achievement.**

Hope Can Be Curative.

Podcast episode of *Improbable Developments* with **Dr. Dave Pearce**, President of Innovation and Research Sanford Research and and Vice Chair of the IRDiRC Consortium Assembly. Dr. Pearce shares his inspiring story and how he got involved in Batten Disease research and where that work has taken him since.

The IRDiRC Scientific Secretariat wishes you all a Merry Christmas and Happy New Year!



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