

# IRDIRC JANUARY - FEBRUARY 2017 UPDATE



## 3<sup>rd</sup> IRDiRC Conference *Thank you!*

### The 3rd IRDiRC Conference: working towards new rare disease research goals for the next decade

The 3rd International Rare Diseases Research Consortium (IRDIRC) Conference took place in Paris, France, on February 8-9, 2017. Around 300 participants from around the globe, including academics, researchers, industry leaders, policy makers and patient advocates, attended the meeting to help shape the future of rare diseases research.

The conference provided a unique opportunity for stakeholders active in the field of rare diseases research to reflect on the progress made in the last decade, particularly since the launch of IRDiRC in 2011, and to look forward to the challenges ahead.

Three plenary sessions opened the conference, presenting the history and achievements of IRDiRC, providing a global view of rare disease scientific achievement, and introducing the state of foundational, diagnostics, and therapeutic rare diseases research. In the first series of parallel sessions, the state of foundational, diagnostics and therapeutics research was investigated further, and included various success stories in each space. This was followed by a parallel session on new approaches to rare diseases, which highlighted the innovative contributions of young investigators towards advancing rare disease research goals. The third parallel session was aimed at exploring trends in the fields of regulatory and access, patient advocacy, and companies.

The conference closed with a forward-looking plenary, following the theme of the conference, transforming rare diseases research. In this session, the concepts for the next set of goals for IRDiRC and the rare diseases research community were outlined. A panel discussion followed, in which the audience was invited to comment, discuss, and ask questions, to further shape the vision and objectives for the next decade. An active and dynamic discussion with the audience and panel followed, which ended the lively and interactive 3rd IRDiRC Conference.

The conference was made possible through the generosity and continuous support of the European Commission, Japan Agency of Medical Research and Development, E-Rare and Orphanet. IRDiRC would like to thank all participants for their active involvement, and their continuous support in helping build and transform rare diseases research.

## Upcoming IRDiRC teleconferences and meetings

- March 1, 2017 – Operating Committee – Teleconference

## Research highlights from IRDiRC members

- NCATS celebrates [five years of advancing translational sciences](#)
- Trans-NCATS collaboration enables rapid advancement of [rare lung disease therapy](#) to human trials
- 10 years of [E-Rare video](#)
- NCI new drug formulary will help [expedite use](#) of agents in clinical trials
- NHGRI turns [20!](#)
- [Jeff Schloss](#), NHGRI's catalyst for DNA sequencing technology development, retires
- The next step in studying how [movement becomes medicine](#)
- [FDA Patient Representative Program](#): looking for patients or caregivers
- FDA approves first drug for [spinal muscular atrophy](#)
- [Identification of mutation](#) supported by the French Foundation for rare diseases
- [Lysogene](#) announces the registration of its *document de base* in relation to its planned IPO on Euronext's regulated market in Paris
- Genetic Alliance study is open: Participants are now [recruiting investigators](#)
- [Orphan drugs](#) represent 41 percent of all new medications
- 2017: Looking forward as [EURORDIS](#) celebrates 20 years
- Geneva hosts first [Rare Diseases International](#) policy event

## Research highlights published on the website

- Approval of [European Reference Networks](#)
- The European Parliament calls for a revision of the [Pediatric Medicines Regulation](#)
- [Patient-Reported Outcome](#) labeling in the United States
- Recommendations to return [research participant's genomic results](#) to relatives

## Research highlights

### IRDiRC Consortium Assembly and Scientific Committee meeting 2017:

In mid-February, IRDiRC Scientific Committees and IRDiRC Consortium Assembly meeting was held in Paris, France, prior to the 3rd IRDiRC Conference. These meetings had a dual aim: to discuss the progress that the Consortium Assembly, Scientific Committees, Task Forces and Constituent Committees have made over the past year, and also to work towards shaping and discussing the new set of IRDiRC goals for the next decade. Now that one of IRDiRC's major objectives, i.e. delivering 200 new therapies for rare diseases, has been reached, and the other objective, the means to diagnose most rare diseases, has made significant progress, the time has come to determine the new vision and objectives for 2017-2027.

### Petra Kaufmann elected as new Chair of the Interdisciplinary Scientific Committee

IRDiRC proudly announces that Dr Petra Kaufmann has been elected as Chair of the Interdisciplinary Scientific Committee (ISC), where she will serve with the newly elected Vice Chair Dr Domenica Taruscio. Dr Kaufmann is the Director of the Office of Rare Diseases Research (ORDR), and Director of the Division of Clinical Innovation (DCI) at the National Center for Advancing Translational Sciences (NCATS). Dr Taruscio is a research director of the National Center for Rare Diseases at Istituto Superiore di Sanità

(ISS) and is responsible for the Italian National Registry of Rare Diseases. Dr Kaufmann will follow in the footsteps of Professor Hanns Lochmüller, who is stepping down after having served five years as Chair, but will remain committed to the work of the ISC, and its Task Forces. IRDiRC would like to thank Professor Lochmüller for all his continued efforts as ISC Chair, and is glad that he will remain a valuable member of the Committee.

### **Chairs elected for the IRDiRC Constituent Committees**

In August 2016, three Constituent Committees were created, a Funders Constituent Committee, Companies Constituent Committee and a Patient Advocates Constituent Committee, in order to better coordinate, enhance information exchange, identify common goals, and advance towards IRDiRC objectives within the different constituent groups.

Dr Daria Julkowska, representing ANR, the national French research agency, and Coordinator of E-Rare, has been elected as Chair of the Funders Constituent Committee. She brings with her a wealth of experience concerning the coordination and management of international research funding programs for rare diseases, and the development of partnerships between funding agencies. She will be supported by Professor Hugh Dawkins, Vice-Chair of the Funders Constituent Committee, and Vice Chair of the IRDiRC Consortium Assembly, and Director of the Office of Population Health Genomics (OPHG), Western Australian Department of Health.

Dr David Thompson was elected as Chair of the Companies Constituent Committee. He is Senior Vice President and Global Head of Research and Nonclinical Development at Shire. He has a large experience in the pharmaceutical industry, with various responsibilities, such as the responsibility for initiating new research programs and external partnerships.

Ms Sharon Terry was elected as Chair of the Patient Advocates Constituent Committee, where she will serve with Vice Chair Ms Béatrice de Montleau. Sharon Terry is President and CEO of Genetic Alliance, and founder of PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE), in response to the diagnosis of PXE in her two children in 1994. She has been at the forefront of patient advocacy participation in research for many years. Béatrice de Montleau is a Board Member of EURORDIS-Rare Diseases Europe.

With the election of the Chairs of the Constituent Committees, IRDiRC is confident to have made a step in the right direction, to advance towards the fulfilment of new goals and objectives, and to contribute to the next phase of IRDiRC.

### **Rare diseases training capacity and needs survey 2017 Elixir**

ELIXIR-EXCELERATE prepared a survey to collect training capacity and needs of the rare disease community across Europe. This will enable better understanding of the current training landscape in rare diseases across ELIXIR nodes or wider across the rare diseases community.

ELIXIR kindly asks you to complete the survey and to forward it to any other interested parties across the rare diseases community: <https://www.surveymonkey.com/r/GQ5M68X>

## **IRDiRC-related calls**

The Canadian Institutes of Health Research has launched a call entitled '[Joint Programme on Neurodegenerative Disease Research \(JPND\) 2017](#).' This call is aimed at to encourage combined analysis of neurodegenerative diseases across traditional clinical boundaries, technologies and disciplines. Application deadline: March 6, 2017.

## **Other news**

If you are presenting in a meeting or a conference, and you would like to show some information about IRDiRC, standard slides are available on the IRDiRC private website; additional slides can be made

available upon request. Please also email the Scientific Secretariat when and where you will be presenting, so we can keep track of "IRDiRC presence at conferences."

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