

# IRDiRC JULY-AUGUST 2016 UPDATE



## Save the date for the 3rd IRDiRC Conference in Paris, France

The third conference of the International Rare Diseases Research Consortium (IRDiRC) will take place February 8-9, 2017 in Paris, France at the conference center of Université Pierre et Marie Curie (UPMC) Jussieu.

Five years have passed since the initial launch of IRDiRC in 2011 and considerable advances have been made in rare diseases research. This conference presents a unique opportunity to reflect upon this progress, confront barriers, and together, form new collaborations to take on challenges posed by rare diseases. Past conferences were held in Dublin, Ireland and Shenzhen, China.

All stakeholders – investigators, policy makers, opinion leaders, critical thinkers, young researchers and patient advocates alike – active in the area of rare diseases from across the globe are invited to join us to celebrate achievements in the field, identify future milestones and goals, and work toward bringing diagnoses and therapies to all rare disease patients.

Registration for this conference will open shortly.

## Upcoming IRDiRC teleconferences and meetings

- September 1, 2016 – Conference Planning Committee – Teleconference call
- September 2, 2016 – Participant Unique Identifiers Task Force – Teleconference call
- September 7, 2016 – Operating Committee – Teleconference call
- September 22-23, 2016 – Consortium Assembly – Face-to-face meeting, Catania, Italy
- September 27, 2016 – Data Mining and Repurposing Task Force – Teleconference call
- November 16, 2016 – Data Mining and Repurposing Task Force – Workshop, Barcelona, Spain
- December 8-9, 2016 – Participant Unique Identifiers Task Force – Workshop, Paris, France
- February 6-7 – Consortium Assembly and Scientific Committees – Face-to-face meeting, Paris, France
- February 8-9, 2017 – 3rd IRDiRC Conference - Paris, France

## IRDiRC new documents

- Consortium Assembly – [Report](#) of the teleconference meeting held on August 2, 2016
- Small Population Clinical Trials Task Force – Workshop [report](#) and recommendations

## Rare disease research published on the website

- Four applicants to benefit from the [PRIME initiative](#) by the EMA
- Statistical design and analysis of [small population group trials](#)
- Worldwide collaboration for [orphan drug designation](#)
- Report on the multi stakeholder for regulation of [advanced therapy medicines](#)
- Open for public consultation: [Innovative Medicines Initiative draft report](#) on facilitating the translation of advanced therapies to patients in Europe
- FDA requests comments on draft guidance for [Precision Medicine Initiative](#)

## Research highlights from IRDiRC members

- Preliminary results reporting the [safety and effectiveness of gene therapy](#) in patients with metachromatic leukodystrophy published in The Lancet
- [Progeria](#) cure remains elusive but new therapeutic options are emerging
- Researchers advance treatment possibilities for [Gaucher](#), Parkinson's
- The power of model organisms for [studying rare diseases](#)
- NIH awards \$55 million to build million-person [precision medicine study](#)
- [The China Precision Medicine Cloud](#): A world-leading platform to benefit patients and health
- Significant expansion of data available in the [Genomic Data Commons](#)

## Research highlights

### Recommendations for the Design of Small Population Clinical Trials

The IRDiRC Small Population Clinical Trials (SPCT) [Task Force](#) has published its [report](#), which includes recommendations and guidelines for the design of small population clinical trials in the field of rare diseases, resulted from the IRDiRC SPCT workshop that was held at the EMA premises on March 3, 2016 in London, UK.

The Task Force members discussed technical solutions to make the best use of scarce data in the context of small studies, typically in rare diseases, and to identify further areas where research is needed. These recommendations have been set up in order to contribute to ensuring, ultimately, effective therapies for patients. The recommendations are intended to be of guidance to clinicians, researchers and regulators.

Overall, it was concluded that randomized clinical trial is considered the traditional golden standard, but it is strongly advised to look systematically at alternative design options when setting up a clinical trial for a rare disease. Not every rare disease trial is as challenging as others, but if a randomized control design is not feasible, other trial options should be considered. Also, a strong emphasis was put on the promotion of the use of scientific advice from regulators regarding small population clinical trials.

### Creation of Constituent Committees to advise the IRDiRC Consortium Assembly

The International Rare Disease Research Consortium (IRDiRC) has grown substantially over the years, since its launch in 2011. In order to better accommodate the growth and diversity of the members, the IRDiRC governance is currently undergoing a number of changes. While the coordination of science and knowledge sharing among the scientists are central to IRDiRC goals, so are the coordination of work and knowledge sharing for the funders, companies, and patient advocacy groups which make up the Consortium Assembly. Until present, no formal forums exist for the latter.

To this end, three Constituent Committees have been created: Funders Committee, Companies Committee, and Patient Advocates Committee. Members of each committee comprise of representative members of the Consortium Assembly. Nonetheless, each committee will meet and discuss independently of the Consortium Assembly, and provide feedback and advice in a similar manner to that of the Scientific Committees.



The Funders Committee gathers IRDiRC public funders to identify and address gaps in rare diseases research funding, including coordination of funding efforts and new network opportunities. The Companies Committee discusses the collective view from the industry and investigate roadblocks that hold back progress in rare diseases clinical research relative to IRDiRC goals. The Patient Advocates Committee represents patients' needs and look into patient engagement in rare diseases research. All together, the Constituent Committees will identify common priorities and goals, address roadblocks and coordinate solutions, in order to directly advance IRDiRC objectives.

With the creation of these Constituent Committees, IRDiRC is confident that the consortium will continue to grow and thrive, to not only have the structure to fulfill the original goals and objectives but to build towards new ones continuously and continually, and together, contribute to the next phase of IRDiRC.



## IRDiRC's new member

Roche, a Swiss multinational pharmaceutical company, has joined IRDiRC as a new industrial partner. Motivated to contribute to IRDiRC goals and objectives, specifically by the opportunity to provide input in discussions around translational, pre-translational, pre-clinical and clinical research. IRDiRC warmly welcomes Roche and looks forward to a fruitful collaboration.

## IRDiRC-related calls

NORD has launched a call for U.S. and international researchers interested in studying rare diseases. [2016 funding](#) is available for the following diseases: Alveolar Capillary Dysplasia, Appendix Cancer and Pseudomyxoma Peritonei, Autoimmune Polyglandular syndrome Type 1, Homocystinuria, Malonic Aciduria and Stiff Person syndrome. Application deadline: September 12, 2016.

The French Foundation for Rare Diseases has launched a call entitled "[GenOmics of rare diseases.](#)" The goal of the call is to support hypotheses driven research projects aimed at exploring genetic and molecular bases of rare diseases by the use of next generation sequencing approaches to make progress in the understanding of rare diseases with the aim to improve therapeutic strategies. Application deadline: September 27, 2016.

## 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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