

# IRDIRC SEPTEMBER-OCTOBER 2016 UPDATE



## Registration is open for the 3rd IRDiRC Conference in Paris, France

We invite you to join us at the 3rd conference of the International Rare Diseases Research Consortium (IRDIRC), which will take place February 8-9, 2017 in Paris, France at the conference center of Université Pierre et Marie Curie (UPMC) Jussieu.

A strong and exciting program has been put together, with 4 plenary and 3 parallel sessions, featuring prominent international speakers. At a glance, the program will start by discussing the state of rare diseases research on a global scale, and will be followed by highlights of transformative efforts within the areas of foundational, diagnostics and therapeutics research. On the second day, new approaches to rare diseases research and trends in the field will be highlighted. The conference will close by discussing how to transform rare diseases research over the course of the next 10 years, including a panel discussion on the next set of IRDiRC goals for 2017-2027.

In addition to compelling talks, the 3rd IRDiRC Conference offers an array of networking opportunities for a diverse set of rare disease professionals and stakeholders from the rare disease world. Young investigators are encouraged to submit abstracts for oral and poster presentations.

Registration and abstract submission is now open, and the early registration rate is in effect until November 30, 2016. Reduced rates are available for trainees, patient advocates, and IRDiRC Scientific Committee and Task Force members.

For more information and registration, go to [www.irdirc-conference.org](http://www.irdirc-conference.org).

## Upcoming IRDiRC teleconferences and meetings

- 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, 2017 – Consortium Assembly and Scientific Committees – Face-to-face meeting, Paris, France
- February 8-9, 2017 – 3rd IRDiRC Conference - Paris, France

## Rare disease research published on the website

- Rethinking [variant information](#) linked to rare diseases
- The ethical debate: [de-identification and re-identification](#) of individuals in rare disease research
- [Patients as key partners](#) in rare disease drug development
- [The expectations](#) of rare disease patients on large-scale data sharing for genomics research
- Report: United Nations Secretary-General's High-level panel on [access to medicines](#)

## Research highlights from IRDiRC members

- AFM-Telethon highlights the 30th anniversary of the [DMD gene](#) discovery
- NCATS to fund the next phase of [tissue chip for drug screening](#) program
- From [epigenetics](#) - new molecular switches to silence genes
- Gene therapy shows promise for treating [Niemann-Pick disease](#) type C1
- PTC' [Spinal Muscular Atrophy](#) program advances into Phase 2 clinical studies in SMA patients with RG7916
- Nominations open for [NORD's 2017 Rare Impact Awards](#)
- Genetic Alliance supports the [new NIH Policy on clinical trials](#)

## Research highlights

### 'IRDiRC Recognized Resources': a new mechanism to support scientists to conduct efficient, high-quality research for rare diseases

An [article](#) on 'IRDiRC Recognized Resources' is published in the European Journal of Human Genetics (EJHG); the article reviews 13 different resources that have successfully obtained this quality label in the first year of its implementation. These included data sharing for discovery, knowledge organization and ontologies, networking patient registries, and therapeutic development.

### Guidelines for the informed consent process in international collaborative rare disease research receives the 'IRDiRC Recognized Resources' label

On September 7, 2016, the '[Guidelines for the informed consent process in international collaborative rare disease research](#)' received the 'IRDiRC Recognized Resources' label. The Guidelines are intended to provide guidance for an effective informed consent process focusing on rare disease patients involved in international research. The article that sets out the Guidelines provides an excellent overview of the need for the development of a set of core elements that will allow the best possible informed consent documents for studies and research activities going on in international consortia.

### Draft version of ADA-Matrix was open for comments

The Automatable Discovery and Access (ADA) Task Force has completed a first version of the ADA Matrix and supporting documents. The ADA Matrix (ADA-M), provides a standardized way to represent any and all consent and other conditions of use that apply to a resource, making such information unambiguous, computer-readable and hence directly available for digital communication, searching and automation activities. Making use of ADA-M is expected to reduce the costs and burdens of data stewardship, while increasing the efficiency of responsible data access (discovery and sharing). This draft version was open for comments, and the Task Force currently works on furthering the ADA-Matrix.

### Few patients, but strong evidence: Small Population Clinical Trials Task Force in the spotlight

An article in [DIA's Global Forum](#), the Small Population Clinical Trials (SPCT) Task Force and its resulting report were put in the spotlight. In a special section dedicated to rare diseases, Simon Day, Chair of the SPCT Task Force, sets out the different conclusions and recommendations that were reached at the Task Force workshop held in March 2016.

## IRDiRC-related calls

AFM-Telethon has launched a call entitled "[9th Call for SMA Research Proposals](#)." This new Call for SMA Projects is open to any research project (those of a collaborative nature are encouraged) aimed at finding a therapy for Spinal Muscular Atrophy (SMA) or at elucidating the basic pathophysiological processes of the disease. Application deadline: December 8, 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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