

# IRDiRC MARCH 2016 UPDATE

## Small Population Clinical Trials Task Force workshop



A workshop on [Small Population Clinical Trials](#) took place on March 3, 2016. This workshop, jointly organized by the European Medicine Agency (EMA) and the International Rare Disease Research Consortium (IRDiRC), was held at the EMA premises in London, UK. The purpose of this workshop was to advance technical solutions to make the best use of scarce clinical data in the context of small population trials and to identify points of agreement between the different stakeholders regarding non-classical designs. The workshop was also aimed at identifying further areas where research is needed. Furthermore, its purpose was to issue recommendations that are useful to clinicians and researchers when planning trials.

The workshop, chaired by Dr. Simon Day and Dr. Kristina Larsson, started with a general discussion on the topic, followed by two breakout sessions on six topics: a matrix of the different study methods, obtaining adequate safety data, multi-arm designs, decision analytic approaches, extrapolation and patient' engagement. The day was concluded in a joint session, in which participants gave feedback from their breakout sessions, and discussed and agreed on proposed conclusions and recommendations.

## Rare disease events

### International Congress of Human Genetics 2016

From April 3, 2016 till April 7, 2016, the 13th International Congress of Human Genetics ([ICHG2016](#)) will take place in Kyoto, Japan. The ICHG meeting has been held every five years since 1956, the inaugural in Copenhagen, Denmark, and will be organized for the first time in Asia. ICHG2016 will be an exciting opportunity to share latest development in human genetics and discuss the future of human genetics. On April 6, 2016, IRDiRC, together with the Global Alliance for Genomics and Health ([GA4GH](#)), will come together at a Special Focus Session, entitled "IRDiRC, GA4GH and Matchmaker Exchange."

ICHG2016, April 3-7, 2016, Kyoto International Conference Center - Kyoto, Japan

More details: <http://www.ichg2016.org/>

### H2020 European Health/Rare Diseases Brokerage Event – Oslo 2016

The Research Council of Norway, the French Institutes / Embassies in the Nordic countries, and the French Norwegian Foundation will host the next Horizon 2020 Health Brokerage Event in Oslo on rare diseases on May 12-13, 2016. For universities, research institutions, hospitals, companies and patient organizations that intend to prepare a proposal for the challenge in Horizon 2020, this day will present them with more information about the new Horizon 2020 calls dedicated to rare diseases. In addition, there is also a chance for participants to present their projects and to meet matching partners to advance their projects.

H2020 European Health/Rare Disease Brokerage Event, May 12-13, 2016, Research Council of Norway - Oslo, Norway

More details: <https://www.b2match.eu/h2020oslo2016>

## Upcoming IRDiRC teleconferences and meetings

- April 6, 2016 – Operating Committee – Teleconference call
- April 14, 2016 – Participant Unique Identifiers Task Force – Teleconference call
- April 26, 2016 – Data-Mining and Repurposing Task Force – Teleconference call Steering Committee

## Rare disease research published on the website

- Utility of [MAST](#) in European patient-centered telemedicine pilots
- [Expanded access](#) for experimental drugs in the United States: a discussion
- Open for [public consultation](#): Principles for value assessment and funding processes for orphan medicinal products
- [Patenting](#) next generation sequencing technologies: questions and concerns
- A rare disease strategy for [Canada](#)

## Research highlights from IRDiRC members

- [NIH](#) of the United States includes rare diseases in its strategic plan for fiscal years 2016-2020
- [NORD-FDA](#) collaborate to collect natural history data on rare diseases
- Gene therapy: The view from [NCATS](#)
- [NICHD](#) invites researchers to share their data through online resource
- [Lysogene](#) to host first-ever research symposium dedicated to GM-1 Gangliosidosis
- [Sanofi Genzyme](#) leadership in rare diseases highlighted at WORLDSymposium 2016
- Dr. Odile Boespflug-Tanguy, new president of the [AFM-Téléthon](#) Scientific Council
- Future directions for Undiagnosed Diseases Research: [UDN](#) and beyond
- The final result of [Telethon](#) 2015 came up to 93850778€!
- [EURORDIS](#) Awards 2016 – winners announced!
- Huawei and [WuXi](#) AppTec join forces to create China Precision Medicine Cloud Platform



## Research news

### Rare Disease Meetings Week in Barcelona, Spain

In the week of March 7 till March 13, Barcelona saw a week full of rare disease events, putting the spotlight on rare disease research. Several international meetings and events were organized: the membership meetings of [E-Rare](#), [Elixir](#), [NeurOmics](#) and [RD-Connect](#); the launch of [Hipbi-RD](#); and the [RE\(ACT\)](#) Congress. Several members of IRDiRC Executive and Scientific Committees participated in these meetings.

Prof. Hugh Dawkins, Vice-Chair of IRDiRC Executive Committee, presented an update on IRDiRC and our global commitment to rare diseases in the NeurOmics2016 meeting. Prof. Hanns Lochmüller also gave an update on IRDiRC at the RD-Connect meeting, indicating that "IRDiRC will meet the goal of 200 new marketed orphan medical products well before 2020."

At the occasion of the RE(ACT) Congress, Dr. Christopher Austin, Chair of IRDiRC Executive Committee, and Mr. Yann Le Cam, Chair of IRDiRC Therapies Scientific Committee, gave the opening speeches. In Dr. Austin's speech, entitled "*Catalyzing Translational Innovation*," he emphasized that "translation in rare disease research is obligatorily a team sport." Mr. Le Cam's speech, "*ReAct for Patient Access to Innovation*", highlighted the bridges that need to be built, to shape the rare disease ecosystem.

## IRDIRC-related calls

The NIH has launched a call for "[Discovery of the Genetic Basis of Childhood Cancers and of Structural Birth Defects: Gabriella Miller Kids First Pediatric Research Program](#)." In this call, the NIH invites applicants to use whole genome sequencing at a Kids First-supported sequencing center to elucidate the genetic contribution to childhood cancers, and to investigate the genetic etiology of structural birth defects. Letter of intent deadline: May 17, 2016.

The European Commission (EC) has launched a call entitled "[European Reference Networks \(ERN\)](#)." An ERN has to fulfill a list of basic requirements which are listed in the Commission Delegated and Implementing Decisions. Expected benefits to patients and healthcare systems include improvements in services delivery, working systems, patient pathways, clinical tools and earlier adoption of scientific evidence. The objective of this call is to provide financial support to ERNs, once they have been established; therefore, only approved ERNs are eligible for co-funding. Application deadline: June 21, 2016.

The NIH has also launched the next "[NeuroNEXT Clinical Trials](#)" funding call. In this call, applications for exploratory clinical trials of investigational agents (drugs, biologics, surgical therapies or devices) that may contribute to the justification for and provide the data required for designing a future trial, for biomarker validation studies, or for proof of mechanism clinical studies are encouraged. Application deadline: August 2, 2016.

All calls can be found on the [IRDiRC-related calls](#) page.

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