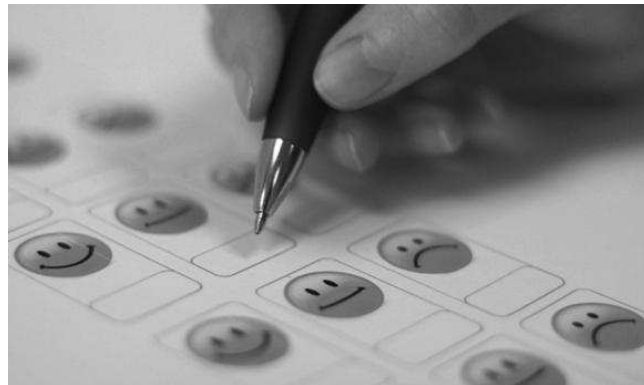


IRDiRC OCTOBER 2015 UPDATE for Executive Committee, Scientific Committees and Working Groups



Draft document on “Patient-Centered Outcome Measures Initiatives in the Field of Rare Diseases” is open for comments

IRDiRC’s Patient-Centered Outcome Measures Task Force will meet at a workshop on November 30, 2015 to identify what should and can be done to support the development of patient-centered outcome measures for rare diseases, in order to improve the feasibility and quality of forthcoming trials and to provide data of relevance to the patient community and other decision makers. A background [document](#) is available, which is currently open for comments and suggestion for the community at large. Please submit comments before November 15, 2015.



Events for your calendar

RE(ACT) Congress 2016

RE(ACT) announces its 3rd international congress on research of rare and orphan diseases. This congress, jointly organized by the Blackswan Foundation and E-Rare, and co-sponsored by IRDiRC, will explore issues and cutting-edge technologies that affect many adult and pediatric conditions. The Congress will bring together world leaders and young scientists from stem cell, cell biology, gene therapy, human genetics and therapeutic applications to present state-of-the-art research, to discuss results and to exchange ideas.

RE(ACT) 2016, March 9-12, 2016, Crowne Plaza Barcelona – Fira Center, Barcelona, Spain

More details and registration: <http://www.react-congress.org/>

Genomics of Rare Diseases: Beyond the Exome

The Genomics of Rare Diseases: Beyond the Exome conference will be held on April 13-15, 2016. This conference, for both clinicians and scientists, will present a blend of genomic science and clinical medicine. The meeting features the latest findings related to the genomic basis of rare diseases, which can provide powerful insights into human biology and focuses on the links between monogenic and polygenic disorders and will explore the various modifiers of rare disease. New technologies to therapy,

including metabolomic approaches, will be discussed. Additionally, discussion will be organized around current ethical issues in genomic medicine.

Genomics of Rare Diseases: Beyond the Exome, April 13-15, 2016, Wellcome Genome Campus, Hinxton, Cambridge, UK

More details and registration: <https://registration.hinxton.wellcome.ac.uk/events/item.aspx?e=575>

Myology 2016

The Fifth International Congress of Myology (Myology 2016), will be held from March 14-18, 2016 in the Lyon Conference Center. Myology 2016 will present multiple pathophysiological and therapeutic breakthroughs and thriving clinical trials in the field. The best specialists in this emerging discipline from the five continents are expected to present and challenge their latest findings not only in fundamental research but also in clinical science and therapeutics.

Myology 2016, 14-18 March 2016, Lyon, France

More details and registration: <http://www.myology2016.org/en/>

IRDIRC new documents

- Executive Committee- [report](#) of the 17th Executive Committee meeting, Montreal, Canada
- Patient-Centered Outcome Measures background [document](#)

Past IRDiRC workshops

- October 6, 2015 - Matchmaker Exchange Task Force- Workshop ASHG 2015, Baltimore, USA
- October 7, 2015 - Matchmaker Exchange Community Engagement Event ASHG 2015, Baltimore, USA

Upcoming IRDiRC workshops

- November 9-10, 2015 - Machine Readable Consent Task Force - Workshop, Paris, France
- November 30, 2015 - Patient-Centered Outcome Measures Task Force - Workshop, Paris, France

Upcoming IRDiRC teleconferences and meetings

- November 12, 2015 – Operating Committee – teleconference call (every second Thursday the month, until March 2016)
- March 14, 2015, Morning – Individual Scientific Committee meetings – Lyon, Paris, France
- March 14, 2015, Afternoon – Joint Scientific and Executive Committees meeting – Lyon, Paris, France
- March 15, 2015 – Executive Committee meeting – Lyon, Paris, France

Research highlights published on the website

- How do [pediatric biobanks](#) look at various aspects of obtaining consent from the pediatric population
- [Long tail economic](#)s and rare disease research: impact of next generation sequencing for rare Mendelian disorders
- [AFM-Téléthon](#) launches a pharmaceutical development center for the production of gene and cell therapies
- [Matchmaker Exchange](#): building evidence for candidate genes
- Prof. Marc Tardieu appointed Director of [French Foundation of Rare Diseases](#)
- [ADAPT SMART](#): a platform for coordinating Medicines Adaptive Pathways to Patients
- Creating a [sustainable environment](#) for orphan drug development
- Analyzing the ability of fulfilling the [obligations of conditionally-approved drugs](#) in Europe

Research News

ORDO and PhenomeCentral receive the “IRDIRC Recommended” label



On October 8, the Orphanet Rare Disease Ontology (ORDO) and PhenomeCentral both received their “IRDIRC Recommended” status. ORDO, a joint initiative between Orphanet and EBI, provides a structured vocabulary for rare diseases capturing relationships between diseases, genes and other relevant features. It integrates nosology, relationships, databases, classifications and connections with other terminologies. PhenomeCentral, a repository for secure data sharing targeted to clinicians and scientists working in the rare disease community, was developed to facilitate the discovery of multiple individuals affected by the same unnamed disorder across partners around the world.

Currently an additional seven applications for "IRDIRC Recommended" are under review, for which a decision is expected in November.



IRDIRC well-received at the Discoveries & Innovations on Orphan Drugs and ICORD conference in Mexico

Mid October, Mexico celebrated its Global Rare Diseases Week 2015, to put the spotlight on orphan diseases in Latin and South America. The week started with the 4th Latin American Meeting of Rare Diseases on October 12, followed by the Discoveries and Innovations in Orphan Drugs Congress on October 13-14, and ended with the 10th International Conference on Rare Diseases & Orphan Drugs (ICORD 2015) on October 15-16. At the occasion of the Discoveries and Innovation in Orphan Drugs Congress, Dr Ségolène Aymé, Coordinator of the IRDiRC Scientific Secretariat, gave an overview presentation of the [perspective of IRDiRC](#) on the trends and opportunities in the field of orphan drugs research and development. In this presentation, the blooming field of research in rare diseases and the development of orphan drugs were discussed. The audience was reminded to look at the essential and long term common interest at stake across patients, companies and competent authorities, rather than antagonizing the short-term and short-take of diverging interest. A short presentation of [IRDIRC's achievements to date](#) was also given at the ICORD conference. This presentation concluded with a future perspective focusing on the different IRDiRC Task Forces.

Paul Lasko interviewed at the ASHG 2015 on IRDiRC and the Matchmaker Exchange



At the annual American Society of Human Genetics congress (ASHG 2015), Professor Paul Lasko, Chair of IRDiRC, was interviewed about [IRDIRC goals and achievements](#). He also spoke briefly about the [Matchmaker Exchange](#) initiative, highlighting the recent developments.

Matchmaker Exchange at ASHG 2015

At the ASHG 2015, two workshops were co-organized by IRDiRC for Matchmaker Exchange. On 6 October 2015, a full-day meeting with different experts was held. The workshop was organized in three parts: the morning was dedicated to meeting with the existing Matchmaker Exchange services; this was flawlessly followed by a discussion to expand the Matchmaker Exchange platform, in which several dedicated new matching services were invited; the day was terminated by a debate about the future and current status of Matchmaker Exchange. On 7 October 2015, a community engagement event was organized, in which about 80 participants were present to learn the ins and outs of this tool.

IRDIRC-related calls

The FDA has launched a research funding initiative for Clinical Studies of Safety and Effectiveness of Orphan Products. The [grants](#) are available to any foreign or domestic, public or private, for-profit or non-profit entity.

The FDA has also launched a call for investigator initiated multi-site clinical trials. [Grants](#) are available for both rare and common disease trials.

AFM-Téléthon has launched an international [call](#) for Strategic Myotonic Dystrophy (DM) Translational Research Projects, to promote collaborative and translational research activity in the field of myotonic dystrophy.

The NIH has launched a [call](#) for pre-clinical research based on existing repurposing tools. This call support rigorous, pre-clinical studies that establish the rationale for a clinical trial, where the hypothesis originates from use of a published or publicly available method for identifying new indications for existing drugs.

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



Other News

IRDiRC on Twitter

Opinions about social media are diverse, but IRDiRC has finally made the leap and decided to join Twitter in September. So for those that like their fast regular dose of IRDiRC and rare disease news, follow our account [@irdirc](#).

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<http://www.irdirc.org/>