

IRDiRC MAY 2015 UPDATE

for Executive Committee, Scientific Committees and Working Groups



Launch of IRDiRC Task Forces

Matchmaker Exchange Task Force has been launched and is holding its first discussions and workshop.

A core group has now been constituted for Patient Relevant/Reported Outcome Measures Task Force and is ready to discuss the final composition of the Task Force. The group's first conference call is scheduled for 15th June.

Core members of Small Population Clinical Trials and of Data Mining & Repurposing Task Forces have been nominated and will hold their first discussions over the coming weeks to enlarge the Task Forces' membership and define their strategy.

IRDiRC new documents

- **Executive Committee:** [report](#) of the teleconference meeting held on May 11, 2015.
- **Diagnostics Scientific Committee:** [report](#) of the teleconference held on May 12, 2015.
- **Interdisciplinary Scientific Committee:** [report](#) of the teleconference held on May 15, 2015.

Upcoming IRDiRC meetings and teleconferences

- **June 5, 2015:** meeting of the Diagnostics Scientific Committee in Glasgow, UK (full day)
- **June 5, 2015:** meeting of the Interdisciplinary Scientific Committee in Glasgow, UK (full day)
- **June 5, 2015:** meeting of the Therapies Scientific Committee in Glasgow, UK (afternoon)
- **June 6, 2015:** meeting of the three Scientific Committees in Glasgow, UK (morning)
- **August 4, 2015 – Executive Committee meeting – teleconference**
- **September 11, 2015:** Executive Committee meeting in Montreal, Canada

Research highlights published on the website

- The [TREAT-NMD advisory committee](#) for therapeutics and its role in bringing orphan drugs to the market
- [Anonymising and sharing individual patient data](#)
- Identification of a large [set of rare complete human knockouts](#)
- Scientists' perspectives on [consent in the context of biobanking research](#)
- Scientific advice leads to [stronger applications from industry](#)
- The [discovery of medicines for rare diseases](#)
- [WHO issues statement to support clinical trial reporting within 12 months of completion](#)

- Recommendation for a [pan-European registry of paediatric cancers](#)
- Factors influencing [drug development for lysosomal storage disorders](#) in the United States
- The [Canadian Open Genetics Repository](#) for clinical genomics

IRDiRC-related calls

- The Canadian “[Rare Diseases: Models & Mechanisms](#)” Network (RDMM)'s call for applications has been added to the [IRDiRC-related calls](#) page.

Website updates

- The [IRDiRC presence at conferences section](#) has been updated with the [Education Session \(134\) on Orphan Diseases](#) during the ASGCT 18th Annual Meeting which took place in New Orleans, USA, from 13 to 16 May 2015. Ségolène Aymé presented the challenges of orphan drug development and solutions identified by IRDiRC to overcome or minimise these obstacles.

IRDiRC in the News

- *Disease Models & Mechanisms* published an interview with Kym Boycott on May 1, 2015 (8:417-419): [Looking for a needle in a haystack – tackling rare diseases](#). Kym talks about her career as clinical geneticist at the Children's Hospital of Eastern Ontario (CHEO) and her team's work aiming to bridge basic and clinical knowledge to improve diagnosis and management of rare diseases. As co-leader of the Canadian Rare Diseases Models and Mechanisms (RDMM) project, Kym aims to help understand disease mechanisms and facilitate therapeutic configuration for rare diseases.
- The French journal [Pharmaceutiques](#) published an issue on April 1, 2015 (in French), focusing on rare disease research in France. A section on IRDiRC discusses some of the consortium's ongoing activities, such as IRDiRC's ambition to include patient organisations into clinical research protocols, establish a consensus on methods to conduct small population

clinical trials, and make sequencing databases and clinical data interoperable.

Other News

Sophie Höhn, former communication manager at IRDiRC Scientific Secretariat, has been hired by Orphanet to work on the scientific content of OrphaNews. We wish her all the very best in this demanding and exciting activity.

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