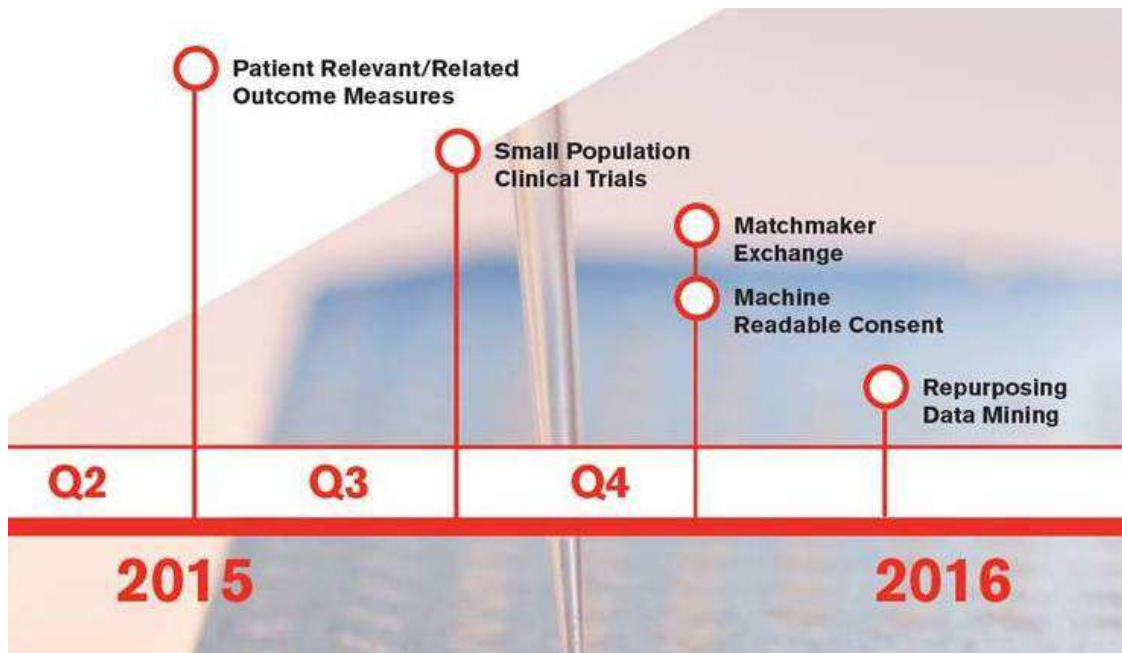


# IRDiRC APRIL 2015 UPDATE

## for Executive Committee, Scientific Committees and Working Groups



### IRDiRC 2015-2016 Activity Plan

Task Forces are being constituted around the following selected projects to organise workshops, and produce, disseminate and implement recommendations resulting from these session outcomes.

- Patient Relevant/Reported Outcome Measures
- Small Population Clinical Trials
- Matchmaker Exchange
- Machine Readable Consent
- Data Mining & Repurposing

All workshops will come under the umbrella of IRDiRC and will not be exclusively attached to a specific Scientific Committee, in order to optimally benefit from the extensive clinical and scientific expertise within the three committees.

Members of Task Forces are being nominated based on their specific expertise in the selected fields. They will include key players from different backgrounds (e.g. academia, industry, regulatory, advocacy), in order to approach topics from all perspectives and respond to all needs in these fields.

## IRDiRC new documents

- Executive Committee: [report](#) of the meeting held in Madrid, Spain, on March 16, 2015.
- Ontologies Working Group: [report](#) of the teleconference held on March 11, 2015.

## Upcoming IRDiRC meetings and teleconferences

- May 11, 2015: teleconference of the Executive Committee
- May 12, 2015: teleconference of the Diagnostics Scientific Committee
- May 15, 2015: teleconference of the Interdisciplinary Scientific Committee
- 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)
- September 11, 2015: Executive Committee meeting in Montreal, Canada
- April 14-15, 2016: IRDiRC Executive Committee meeting – Windsor, UK

## Research highlights published on the website

- FDA formalises [expedited review procedures](#) for marketing applications with breakthrough therapy-designated drugs and biologics
- Use of [Electronic Informed Consent in Clinical Investigations](#): guidance document by FDA open for public consultation

- Challenges and bottlenecks for [genomic data sharing](#)
- [CoBRA](#): guidelines to standardise citation of bioresources in journal articles
- Practical guidance on [informed consent for paediatric participants](#) in a biorepository
- Scientific framework to use the [accelerated approval pathway](#) and to qualify biomarkers as primary endpoints to develop orphan drugs

## IRDiRC-related calls

- [AFM Telethon call for proposals](#) in Spinal Muscular Atrophy and Collagen VI projects. International scientific experts are invited to apply.
- NIH funding opportunity announcement for [Validation of Paediatric Patient Reported Outcomes in Chronic Diseases](#)
- NIH funding opportunity announcement for [Undiagnosed Diseases Gene Function Research](#)
- NINDS call for applications for a [Human Biospecimen and Data Repository for Biomarkers Research](#) ("Biomarkers Repository") in Neurological Disorders.

## Website updates

- The [IRDiRC website](#) is undergoing a complete makeover to improve clarity, and highlight essential documents and IRDiRC activities. Thank you for your patience while the new website takes shape.
- The [IRDiRC presence at conferences section](#) has been updated with the addition of a session on "IRDiRC genome and phenome: why data sharing is important for rare disease research" during the conference on [Genotranslation: Interpretation of Genome Data in Diagnostics at the 3Gb-TEST Central and Eastern European Regional Meeting](#) which took place in Prague, Czech Republic, on April 22, 2015. The meeting consisted of a course for 150 participants with lectures and workshops on next-generation-sequencing technologies and variant interpretation for diagnostics, a public symposium on Rare Disease Research and a business meeting of the European project with review of deliverables and milestones and further planning. The final meeting of this 2-year funded project will be held in August 2015 in Leiden, NL.

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