IRDiRC JUNE 2016 UPDATE

IRDiRC's Consortium Assembly at the center of its governance

The International Rare Disease Research Consortium (IRDiRC) was started in 2010 with the objective to accelerate medical breakthroughs for people affected by rare diseases. Initially conceived as a consortium of rare disease research funders following a joint discussion of the European Commission and the US National Institutes of Health, IRDiRC has experienced substantial growth in the number and diversity of its members over the last five years. Current Consortium members include funding agencies, industry, institutes, ministries and patient organizations.

For the Consortium to continue to function efficiently, IRDiRC governance needs to adapt to the growth in size and diversity of its members and functions. The first of these adaptations is the renaming of IRDiRC's largest representative body, formerly the Executive Committee, as the Consortium Assembly. The new name more accurately reflects this group's function as a gathering of all the consortium's members, focused on information exchange and efforts to develop and coordinate scientific and policy efforts that will advance IRDiRC goals.

IRDiRC new documents

• Diagnostic Scientific Committee – report of the11th Diagnostic Scientific Committee meeting – Teleconference
• Executive Committee – report of the 20th Executive Committee meeting – Teleconference

Upcoming IRDiRC teleconferences and meetings

• July 6, 2016 – Operating Committee – Teleconference call
• July 26, 2016 – Data Mining and Repurposing Task Force – Teleconference call
• August 2, 2016 – Consortium Assembly – Teleconference call
• August 17, 2016 – Participant Unique Identifier Task Force – Teleconference call
• September 22-23, 2016 – Executive Committee – Face-to-face meeting, Catania, Italy
• November 16, 2016 – Data Mining and Repurposing Task Force – Workshop, Barcelona, Spain
• December 8-9, 2016 – Participant Unique Identifier Task Force – Workshop, Paris, France

Rare disease research published on the website

• The orphan drug pipeline in Europe
• Small and Medium sized initiative of the EMA celebrates its 10th anniversary
• PhRMA reports on the progress on treatments in rare diseases in the United States
• Rethinking the ethics of Biobanking
• The European Medicines Agency publishes its annual report 2015

Research highlights from IRDiRC members

• Shire completes combination with Baxalta creating the global leader in rare diseases and highly specialized conditions
• European rare disease patients call for increased European collaboration on medicine pricing to improve access

• EURORDIS CEO Yann Le Cam appointed as patient representative to the EMA Board, a tribute to the rare disease community
• Western Australia launches Undiagnosed Disease Program
• The China precision medicine cloud: a world-leading platform to benefit patients and health
• First European market authorization for a gene therapy medication for a immunodeficiency
• NORD appoints genetics and metabolic specialist Marshall L. Summar M.D. as the Chairman of the Board
• NIH finalizes single institutional review board (IRB) policy for multi-site studies

Research highlights

"IRDIRC Recognized Resources" identifies resources for rare disease research communities

The International Rare Disease Research Consortium (IRDiRC) has renamed its quality label, formerly known as "IRDiRC Recommended", to "IRDiRC Recognized Resources."

The new name, "IRDiRC Recognized Resources", better reflects the fact that these are valuable resources – be it a tool, a platform, guidelines or standards – to members of the rare disease community. Resources that have obtained this label underwent a peer-review process by members of the IRDiRC Scientific Committees. Members, often users of these resources themselves, expect "IRDiRC Recognized Resources" to potentially help accelerate the pace of discoveries and translation into clinical applications. We hope the rebranding will give greater clarity to this initiative.

The rebranding is accompanied by a new logo. All new and existing "IRDiRC Recognized Resources" should make the new logo visible on the resource, giving users an assurance of its relevance to and use by the rare disease community. The new logo will be shown on all "IRDiRC Recognized Resources."

The initiative, originally launched in March 2015, has been given to thirteen resources to date, including three guidelines, four platforms and two reference databases.

GARD and Orphanet have signed a collaboration agreement to improve information on rare diseases across the seas

The Genetic and Rare Disease Information Center (GARD), part of NCATS, NIH, and Orphanet, an "IRDiRC Recognized Resource," have signed a collaboration agreement. This agreement will portray information from Orphanet to the GARD website , and vice versa, GARD information on the Orphanet site whenever information about a specific disease is missing. Information from Orphanet's "identity card" of rare diseases will also become visible on the GARD website. In order to do so, an alignment of the specific diseases of the respective sites is being performed. Both rare disease portals will thus gain a wealth of information from this mutual exchange, for the benefit of patients and doctors overseas.

IRDiRC-related calls

AFM-Téléthon is pleased to announce that the funding process to support clinical and genetic registries projects has changed. The applications for funding these projects are now to be submitted outside the AFM-Téléthon annual Call for proposals and can be done throughout the year.

All calls can be found on the IRDiRC-related calls page.

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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