IRDiRC APRIL 2016 UPDATE

IRDiRC Executive and Scientific Meeting: brainstorming sessions

IRDiRC Executive and Scientific Committees met in Lyon on March 14 and 15, for a series of Scientific, Joint and Executive Committees meetings. These meetings were dominated by a number of exciting brainstorming sessions. Two topics that particularly stood out were a discussion around IRDiRC's impact after its first five years, and a brainstorming session for the strategic priorities for 2016-2020. Now that one of IRDiRC's major objectives, i.e. delivering 200 new therapies for rare diseases, is within reach this year, the time has come to focus and stretch out to new goals. In order to further develop the new strategies and perspectives, an Executive Committee Perspective Task Force has been set up.

Upcoming IRDiRC teleconferences and meetings

• May 4, 2016 – Operating Committee – Teleconference call
• May 11, 2016 – Diagnostic Scientific Committee – Teleconference call
• May 17, 2016 – Participant Unique Identifiers Task Force – Teleconference call
• May 23, 2016 – Executive Committee – Teleconference call
• May 24, 2016 – Data-Mining and Repurposing Steering Committee – Teleconference call

Rare disease research published on the website

• EMA launches Priority Medicines Scheme
• European Commission's report on the incentives provided for orphan medicinal products in Europe
• The launch of Italy's Telethon Undiagnosed Diseases Program
• The need for a European orphan device directive
• Early access to medicines for patients suffering from rare diseases
• Commission Expert Group on Rare Diseases: Recommendations to support the incorporation of rare diseases into social services and policies
• Draft reflection paper by the EMA on extrapolating data from adults to children to facilitate pediatric medicine development
• EMA's report of the pilot on parallel regulatory-health technology assessment scientific advice
• RARE-Bestpractices: an update

Research highlights from IRDiRC members

• EORTC's precision medicine study brings therapeutic progress to patients with very rare cancers
• E-Rare, 10 years of collaboration to build a transnational research program on rare diseases
• AMEDs on rare and undiagnosed diseases (IRUD)

• NHGRI celebrates National DNA Day with events that promote genomic literacy
• NORD announces 20 rare disease patient groups selected to develop natural history studies as part of FDA cooperative agreement
• EURORDIS launches Rare Barometer Voices to collect experiences of rare disease patients, family members and patient representatives

Research News

TACT, CTSR and OMIM receive the "IRDiRC Recommended" label

Three new applications were approved by the IRDiRC Scientific Committees for the "IRDiRC Recommended" label: the TREAT-NMD Advisory Committee for Therapeutics (TACT), the Care and Trial Site Registry (CTSR) and Online Mendelian Inheritance in Man (OMIM).

TACT is a unique multi-disciplinary international group of academic and industrial drug development experts as well as representatives of patient foundations and institutional governmental scientific research centers. It meets twice a year to review and provide guidance on the translation and development path of therapeutics programs in rare neuromuscular diseases with large unmet need.

CTSR is aimed to help the pharmaceutical industry and clinical investigators select trial sites as well as to help to identify potential partners for upcoming research projects. The CTSR provides information relevant to clinical trials in the field of neuromuscular and neurodegenerative diseases and to the assessment of 'centers of expertise.' The CTSR collects contact data, patient cohorts, availability of diagnostic tools and equipment, personnel and their clinical trial experience, care settings as well as research and education activities of a site. Funded by the European Union NeurOmics project, the CTSR was expanded in 2013 to include neurodegenerative centres and now encompasses data on patient cohorts of 32 rare diseases.

OMIM is a knowledge base of human genes and genetic phenotypes comprised of over 23,000 structured free-text entries and used weekly by 60-100,000 individuals from all over the world. OMIM names new Mendelian diseases and, in general, splits phenotypes on a molecular basis.

The start of the Participant Unique Identifiers Task Force

On Thursday April 16, the Participant Unique Identifiers Task Force took off with its first leadership call. This joint IRDiRC - GA4GH Task Force is set up after the idea of ISC chair Prof Hanns Lochmüller, Dr Domenica Taruscio and Dr Petra Kaufmann, and is chaired by Dr Petra Kaufmann, Prof Bartha Knopper and Dr Dixie Baker. The objective of this Task Force is to develop a guiding policy for the generation of participant-specific identifiers (pseudonyms) that enable data from the same individual be connected across multiple projects without directly revealing the participant's identity. The first Task Force teleconference call will take place on May 17, 2016.

IRDiRC-related calls

The NIH has launched a call for the "Exploratory Grants in Cancer Epidemiology and Genomics Research." The call invites applications for research on cancer epidemiology, genomics, and risk assessment, in particular for rare and understudied cancer. Application deadline: June 16, 2016; October 16, 2016.

AFM-Téléthon has launched an international call for translation research projects, entitled " RNA as therapeutic target or as therapeutic product." This call is launched to promote translational research activity in the field of RNA as therapeutic target or as therapeutic product and help the dedicated international scientific and medical community in its search for innovative therapies. Application deadline: June 28, 2016.

The FDA has launched a call for "Natural History Studies for Rare Disease Product Development: Orphan Products Research Project Grant." This call is intended to support studies that advance rare disease medical product development through characterization of the natural history of rare diseases/conditions, identification of genotypic and phenotypic subpopulations, and development and/or validation of clinical outcome measures, biomarkers and/or companion diagnostics. Letter of Intent deadline: August 31, 2016.

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

Other News

The IRDiRC Scientific Secretariat has seen some changes recently. Dr Ségolène Ayme has previously stepped down as Coordinator of the IRDiRC Scientific Secretariat, and has been replaced by Dr Ana Rath, Director of Orphanet. Ms Anne-Laure Rey has joined the IRDiRC as administrative assistant, replacing Ms Mariane Bellanger.

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