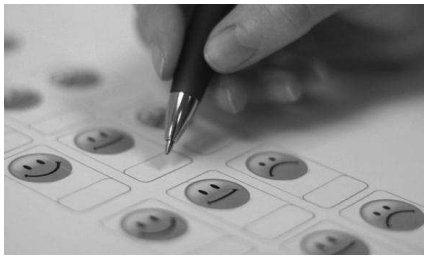
IRDiRC FEBRUARY 2016 UPDATE

Recommendations for the development of patient-centered outcome measures for rare diseases



The IRDiRC Patient-Centered Outcome Measures (PCOM) Task Force has published its post-workshop report, which includes recommendations and guidelines for the development of PCOM. The report and its recommendations have resulted from the IRDiRC PCOM workshop that was organized on November 30, 2015 in Paris, France.

The recommendations cover a large spectrum, and start with recommendations to respect core principles, e.g. clearly defining what, why, how and where to measure, and who is qualified to measure, before the actual measurement. Guidance is provided on the information of what to measure, based on qualitative interviews with stakeholder groups, informing both the benefits and the harms which patients experience.

The recommendations continue with guidance on how to transfer available knowledge on PCOM to the rare disease field, including the preference of adaptation of existing tools to the specificities of a rare diseases. Guidance is given about the search for existing PCOMs, and the assessment of available instruments.

Furthermore, recommendations are outlined to cover the development of new tools as a non-competitive activity. It needs to be taken into account that most rare diseases evolve rapidly over time and therefore require a dynamic model of PCOM. Qualitative research could address critical challenges of outcome measurement in rare diseases, among which: complexity, variability, individualization, lack of background knowledge and small sample sizes.

The recommendations end with guidelines on the sharing of PCOM data and the development of training tools to further inform key actors in the rare disease field.

IRDiRC new documents

• Executive Committee – report of the teleconference meeting held on January 11, 2016
• Interdisciplinary Scientific Committee – report of the teleconference meeting held on February 8, 2016
• Patient-Centered Outcome Measures – post-workshop report and annexes

Upcoming IRDiRC teleconferences and meetings

• March 3, 2016 – Small Population Clinical Trials Task Force – Workshop, London, UK
• March 14, 2016, Morning – Operating Committee meeting – Lyon, France
• March 14, 2016, Morning – Individual Scientific Committee meetings – Lyon, France
• March 14, 2016, Afternoon – Joint Scientific and Executive Committees meeting – Lyon, France
• March 15, 2016 – Executive Committee meeting – Lyon, France
• March 22, 2016 – Data-Mining and Repurposing Task Force – Teleconference

Rare disease research published on the website

• Incorporating patient preferences into drug development and regulatory decision making

• Incorporating patient perspectives in health technology assessments and clinical practice guidelines
• Statement on the utilization of whole-genome sequencing in newborn screening
• Telegenetics in Europe: current usage and constraints

Research highlights from IRDiRC members

• NIH budgets for utilizing genomics to understand rare diseases
• Pooled Health Related Quality of Life Data: large EORTC databases and partnerships demonstrate their value at improving cancer care
• NIH Rare Disease Day event, on leap day (Feb. 29), will feature patients' voices and international collaboration
• Isis Pharmaceuticals changes name to Ionis Pharmaceuticals
• A quarter century after the Human Genome Project's launch: lessons beyond the base pairs
• Research round-up: rare disease research
• #RareDiseaseDay momentum is global and growing!
• Life as a numerator: putting the person in personal genomics

Research News

TREAT-NMD Patient Registries and Standard Operating Procedures receive the "IRDiRC Recommended" label

Two TREAT-NMD resources received the "IRDiRC Recommended" label: TREAT-NMD Patient Registries and Standard Operating Procedures (SOPs) for preclinical efficacy studies.

The TREAT-NMD global network of national registries provides a single entry point for access to patient data worldwide. The TREAT-NMD global registry contains a core set of information including accurate, verified genetic diagnosis together with key clinical data items, all updated at least annually , with submitted data curated and verified at a national level.

SOPs consist of a collection of experimental protocols for the most common outcome measures used to assess drug efficacy in models of neuromuscular disease. The SOPs were developed by a group of preclinical experts and made freely available on the net to facilitate research and as a tool to encourage research groups to choose common procedures in assessing drug or treatment efficacy by different outcome measures.

Testing phase of the Automatable Discovery and Access Task Force tools: the ADA-Matrix

The joint GA4GH-IRDiRC Task Force on Automatable Discovery and Access (formerly Machine Readable Consent) has set out to develop tools to increasingly enable the discovery and access of biomedical resources such as data, biological specimens subjects and software. A key step in this process in the development of an "Automatable Discovery and Access Matrix (ADA-M)", via which consent and other conditions of use can be represented in a standardized, computer-readable manner. ADA-M represents a first draft version of the concept, which is currently looking for Alpha-testers for evaluation and testing. Subsequently, based on the feedback received, the design of the ADA-M will be improved and a related API developed. Both tools will be publicly available via the GA4GH website.

The ADA Task Team is co-chaired by Anthony Brookes (University of Leicester) and John Wilbanks (SAGE Bionetworks), coordinated by Emily Kirby (P3G), and involves over 40 other international experts in data sharing.

IRDiRC-related calls

CIHR has launched a call entitled "Operating Grant : North America Re:Rare." This international program offers the opportunity to develop repurposing projects on proof of concept clinical trials for therapeutic approaches that could be quickly incorporated into clinical practice, and will involve multiple clinical sites located both in Canada and the United States of America.

The French Foundation for Rare Diseases has launched a call entitled "GenOmics: sequencing of rare diseases." The goal of the open call for proposals is to support hypotheses driven research projects aimed at exploring genetic and molecular bases of rare diseases through the use of next generation sequencing approaches to make progress in the understanding of rare diseases with the aim to improve therapeutic strategies.

The European Commission (EC) has launched a call entitled "New therapies for rare diseases". Support will be provided to clinical trials on substances where orphan designation has been given by the EC, where the proposed clinical trial design takes into account recommendations from protocol assistance given by the European Medicines Agency (EMA), and where a clear patient recruitment strategy is presented.

The EC has also launched a call entitled "Diagnostic characterization of rare diseases." The aim of this research call should be to apply genomics and/or other –omics and/or other high-throughput approaches for the molecular characterization of rare diseases in view of developing molecular diagnoses for a large number of undiagnosed rare diseases.

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

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