

IRDiRC JUNE-JULY 2015 UPDATE

for Executive Committee, Scientific Committees and Working Groups



ESHG 2015, Glasgow UK, 6-9 June



IRDiRC held a booth at the European Society of Human Genetics (ESHG) 2015 in Glasgow, UK, from 6th to 9th June, following IRDiRC's individual and joint Scientific Committee face-to-face meetings on 5th and 6th June.

The booth attracted a fair amount of attention and questions from many long-experienced and young budding geneticists.

IRDiRC also presented a poster on the "*Creation of 'IRDiRC Recommended', a label to be used to highlight tools, standards and guidelines contributing to IRDiRC objectives*" in the poster hall.

Launch of IRDiRC Task Forces

The IRDiRC Task Force on Patient-Centred Outcome Measures was launched and its steering committee and members selected among recommended experts in the field of patient outcome measures. Members represent academia, industry and patient organisations.

The workshop will take place in Paris, France, on 30th November 2015.

In the meantime, discussions are ongoing with the steering committee members to finalise the background paper and workshop content.

IRDIRC new documents

- Diagnostics Scientific Committee: [report](#) of the meeting held on June 5, 2015 in Glasgow, UK.
- Interdisciplinary Scientific Committee: [report](#) of the meeting held on June 5, 2015 in Glasgow, UK.
- Therapies Scientific Committee: [report](#) of the meeting held on June 5, 2015 in Glasgow, UK.
- Joint Scientific Committee: [report](#) of the meeting held on June 6, 2015 in Glasgow, UK.

Upcoming IRDiRC meetings and teleconferences

- August 4, 2015 – Executive Committee meeting – teleconference
- September 11, 2015: Executive Committee meeting in Montreal, Canada

Research highlights published on the website

- Outlook of [patient-centered outcomes research](#) in the United States
- [Rethinking patient-reported outcome-based performance measures with the patient in mind](#)
- [CENA approved for \\$1.2 million in second phase of development of PCORnet, a new national clinical research network](#)
- Advantages of comprehensive gene panels as first tier tests over [clinical exome sequencing for Mendelian diseases](#)
- The [European Genome-phenome](#): an EMBL-EBI effort to archive human data consented for biomedical research
- [Nature Genetics urges authors to publish data in managed public repositories](#)

- **The [Human Phenotype Ontology](#): Semantic Unification of Common and Rare Disease**
- **An [algorithm to choose a diagnostic test for Mendelian disorders](#)**
- **Visualising [protein-protein interaction](#)**
- **The [human diseasome](#): phenotype similarity model for common, mendelian, and infectious diseases**
- **Study urges exercising [caution while using variant databases](#) to diagnose rare diseases**
- **[GeneStoryTeller](#): a mobile app for quick and comprehensive information retrieval of human genes**
- **BBMRI-ERIC recommends the [development of biobank-based Expert Centres](#)**
- **[Crowdsourcing](#) project to speed research on rare genetic disorder**
- **[ClinGen](#) — The clinical genome resource of the National Institute of Health in the United States**
- **[CENA approved for \\$1.2 million](#) in second phase of development of PCORnet, a new national clinical research network**
- **Factors influencing [success of clinical genome sequencing](#) across a broad spectrum of disorders**
- **Protein-protein interaction disturbances by missense genetic variants is a very good [predictor of disease severity](#)**
- **A [framework to integrate heterogeneous clinical data](#) into a central registry**
- **[EGI InSPIRE](#) efforts in marking the digital landscape in European Research Area**
- **The [launch of Rare Diseases International](#)**
- **[Drug repositioning](#) can accelerate discovery of pharmacological chaperones**
- **[Randomised trials](#) will serve rare cancers patients better than controlled trials with surrogate endpoints**
- **Now online: updated lists of [medicinal products for rare diseases in Europe](#)**
- **[CORBEL cluster project](#) funded by the European Union**

- **Effective communication between investigators and parents during paediatric clinical trials**
- **Exome sequencing for endocrine disorders: past present and future**
- **Lessons from the Parelsnoer Institute in the Netherlands for biobanking and translational medicine**
- **PharmacoGenomic Mutation Database: a comprehensive manually curated pharmacogenomic database**
- **A Drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases**

IRDiRC-related calls

- The French Fondation Maladie Rare's "High throughput sequencing and rare diseases" Whole Exome Sequencing programme call for applications has been added to the IRDiRC-related calls page.

Other News

IMI has named Pierre Meulien as its new Executive Director. He will take up his duties at IMI in September and brings with him a wealth of experience working in academia, the pharmaceutical industry and research-funding organisations on both sides of the Atlantic. Pierre Meulien is currently President and Chief Executive Officer of Genome Canada, a not-for-profit organisation that connects ideas and people across public and private sectors to find new uses for genomics and invests in large-scale science and technology to fuel innovation. Before joining Genome Canada in 2010, Pierre Meulien served as Chief Scientific Officer for Genome British Columbia, where he promoted the organisation's scientific strategy. Prior to that, he was founding CEO of the Dublin Molecular Medicine Centre (now Molecular Medicine Ireland).

Antonia Mills, IRDiRC Scientific Secretariat, IRDiRC, Plateforme Maladies Rares / Rare Disease Platform, 96 rue Didot, 75014 PARIS, FRANCE, Tel: +33 1 56 53 81 37, Fax: +33 1 56 53 81 38

<http://www.irdirc.org/>

