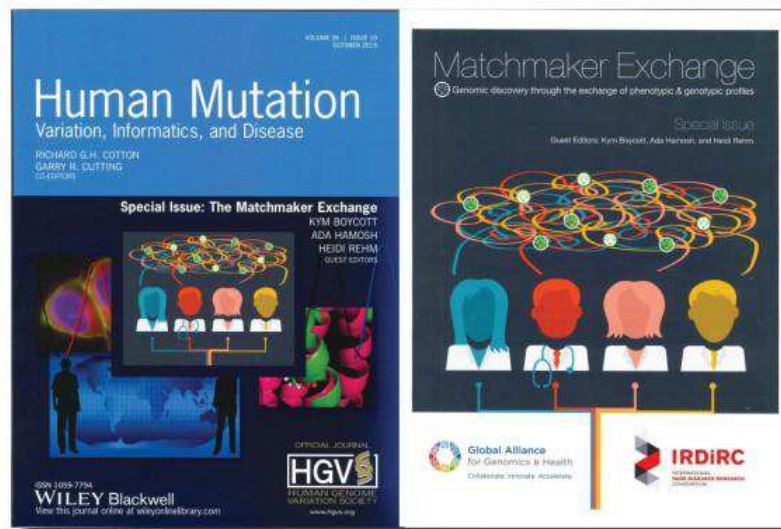


# IRDIRC AUGUST-SEPTEMBER 2015 UPDATE

## for Executive Committee, Scientific Committees and Working Groups



## Special Issue Human Mutation

A special issue of *Human Mutation* covering the Matchmaker Exchange is published. This issue, with guest editors Kym Boycott, Ada Hamosh and Heidi Rehm, covers the overview of Matchmaker Exchange, describes the Matchmaker Exchange API and matchmaking databases, and shares success stories of this initiative. Free copies will be available at the IRDiRC booth at the American Society of Human Genetics conference 2015.



Japan Agency for Medical Research and Development

## IRDIRC's new members

[Pfizer](#), an American multinational pharmaceutical corporation, has joined IRDiRC as a new industrial partner. IRDiRC was also joined by two Japanese agencies, the Japanese [Agency for Medical Research and Development](#) (AMED) and the [National Institutes of Biomedical Innovation, Health and Nutrition](#) (NIBIOHN). IRDiRC warmly welcomes all three new members and looks forward to a fruitful collaboration.

## Events for your calendar

IRDIRC at the ASHG 2015

IRDiRC will be present at the 2015 conference of the [American Society for Human Genetics](#) (ASHG 2015) for a variety of events. IRDiRC will be present at booth (#1904) where everyone can come by for more information and a free copy of the special issue of Human Mutations; co-organise, with GA4GH, a Matchmaker Exchange workshop on 6 October and a community engagement event on 7 October; and present a poster on its Task Forces (506W).



American Society of Human Genetics Annual Meeting 2015, 6-10 October 2015, Baltimore, MD, USA

### **Community Engagement Event Matchmaker Exchange: Building Evidence for Candidate Genes**

Attendees of the ASHG 2015 conference are welcome to join the [Matchmaker Exchange information session](#) to learn more about the platform. Places are limited to 150 places.

Community Engagement Event Matchmaker Exchange, 7 October 2015– 7:00pm - 9:00pm

Baltimore Convention Center – Rooms 339-342, Baltimore, MD, USA

### **MRC organizes a meeting on clinical trials in small populations**

The Medical Research Council's Network of Hubs for Trials Methodology announces a [two-day meeting on clinical trials in small populations](#). The first day will provide an interactive Groundwork Training workshop aimed statisticians and numerate scientists featuring, while the second day will provide a Forward-Looking Forum with invited talks on state-of-the-art methods for trials in small populations.

Clinical trials in small populations: methodological challenges and solutions, 30 November – 1 December 2015, Royal Statistical Society, London, UK

## **IRDiRC new documents**

- Executive Committee- [report](#) of the teleconference call held on 4 August 2015
- State-of-Play 2015 [report](#)

The Executive Committee meeting report will be available in the October newsletter.

## **Upcoming IRDiRC workshops**

- 6-7 October 2015 - Matchmaker Exchange Task Force - Workshop Baltimore, MD, USA
- 9-10 November 2015 - Machine Readable Consent Task Force - Workshop Paris, France
- 30 November 2015 - Patient-Centred Outcome Measures Task Force - Workshop Paris, France

## **Upcoming IRDiRC teleconferences and meetings**

- 8 October 2015 – Operating Committee – teleconference call (this call will be held monthly, every second Thursday the month)
- 14 March 2015 Morning – Individual Scientific Committee meetings – Lyon, Paris, France
- 14 March 2015 Afternoon – Joint Scientific Committee meeting – Lyon, Paris, France
- 15 March 2015 – Executive Committee meeting – Lyon, Paris, France

## **Research highlights published on the website**

- [View](#) of rare disease patients and families on data sharing
- Will [clinical trial data](#) disclosure reduce incentives to develop new uses of drugs?
- The [National Clinical Trials Network](#): conducting successful clinical trials of new therapies for rare cancers
- FDA Guidance document titled "[Rare Diseases: Common issues in Drug Development](#)" for industry is open for public comment

- EMA revised guidelines for [fast track routes](#) for medicines that address unmet medical needs
- [APARDO](#): rare disease alliance for the Asia Pacific region
- [Phenolyzer](#) adeptly prioritises Mendelian and complex disease gene based on free text phenotype searches
- [GeneMatcher](#): a matching tool for connecting investigators with an interest in the same gene
- Targeted next generation sequencing for clinical diagnoses of [561 Mendelian diseases](#)
- [Drug mechanisms](#) with genetic support observe greater success in all phases of drug development

## Research News

### Dr Kaufmann Appointed director of NCATS' Office of Rare Diseases Research

The Office of Rare Diseases Research (ORDR) has named Dr Petra Kaufmann as its new director. She currently directs the National Center for Advancing Translational Sciences (NCATS) Division of Clinical Innovation (DCI) within which the Clinical and Translational Science Awards (CTSA) program resides. From now on, she will direct both the ORDR and the DCI. It is expected that the common leadership for both programs will enhance synergy, increase the centre's ability to advance translational science and help bring more treatments to patients quicker.



### Dr Ségolène Ayme received the insignia of Officer of the Order of the Legion of Honour, France

On 4 September 2015, Dr Ségolène Aymé, coordinator of IRDiRC's scientific secretariat, founder of Orphanet, and Emeritus director of research at INSERM, received the insignia of the Officer of the Order of the Legion of Honour of France for her many contributions to research and patient care for rare diseases. This prestigious declaration was presented to her by Dr Pierre Tambourin, Chief Executive of GIP Genopole Evry, at the platform of rare diseases in Paris, France. In his speech, Dr Pierre Tambourin traced forty years of her professional life dedicated to improving and developing patient care and rare diseases research. He equally referred to her role as women's rights activist and ecologist. Upon accepting the insignia, Dr Ségolène Ayme spoke about the pleasure of speaking the universal language of science, and the privilege to have had the possibility to work internationally over the last twenty years, and she concluded by thanking all past and current collaborative partners for their role in her professional life.

### IRDIRC awarded its first two "IRDIRC Recommended" labels

IRDIRC has awarded its first two "IRDIRC Recommended" labels. "IRDIRC Recommended" is a quality indicator based on a specific set of criteria, aimed to highlight key resources, which would accelerate the pace of discoveries and translation into clinical services. Both the International Charter of Principles for sharing Bio-Specimens and Data, which provides guidance for effective legally- and ethically-grounded sharing of bio-specimens and data and Orphanet, the reference portal for information on rare diseases and orphan drugs, have successfully obtained this quality label.

## IRDIRC-related calls

SMA-Europe has launched a research funding initiative for projects aimed at finding a therapy for [Spinal Muscular Atrophy \(SMA\)](#).

The NIH has launched a research funding initiative for [Polycystic Kidney Disease](#), for both fundamental investigations of the disease and the generation of possible strategies for clinical intervention.

The ANR and the Network of Excellence in Neurodegeneration (CoEN) launched a [Transnational Call](#) aimed at supporting transnational collaborations in the field of neurodegenerative disease.

The NIH announced a funding initiative for institutions proposing research with individuals with [disorders of sex development \(DSD\)](#) and their families.

NINDS has announced a call for [Parkinson's disease \(PD\)](#) biomarker program discovery projects. This call is to study the discovery, assay optimization, and replication stages required for the development of biological biomarkers for Parkinson's disease.

All calls have been added to the [IRDiRC-related calls](#) page.

## Other News

Antonia Mills has left the Scientific Secretariat and Anneliene Jonker has replaced her as Communication Manager.

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