International Rare Diseases Research Consortium Conference 2013

Welcome address

Ruxandra Draghia-Akli, MD, PhD
Chair of IRDiRC's Executive Committee
Director
Directorate for Health
Directorate-General for Research and Innovation
European Commission
Welcome to the first IRDiRC conference

- EU-funded rare disease research in brief
- Introduction to IRDiRC
  - Why work together?
  - Our goals
  - How will it work?
- Some conference information

www.irdirc.org
The EU: A major player in funding health research in rare diseases

- Over two decades of investment in the area
- Close to € 500 million invested in almost 100 projects launched in FP7
- Continued strong investment in Horizon 2020
EU collaborative research in rare diseases

Main focus areas of FP7:

- Europe wide studies of natural history and pathophysiology: in vitro/in vivo models, registries & biobanks, identification of biomarkers
- Development of preventive, diagnostic and therapeutic interventions including pharmacological and innovative approaches
Research challenges related to rare diseases are too big to be mastered alone

- Huge unmet medical needs for patients across the globe
- Small patient populations
- Scarce and scattered research resources and expertise
Why work together?

- Mobilise the necessary critical mass of expertise and resources
- Avoid overlaps in research allowing for more diseases to be tackled
- Deliver new cures and diagnoses to treat patients world-wide
Co-operation at international level to:

- stimulate
- better coordinate
- maximise output

of rare disease research efforts around the world
IRDiRC vision and 2020 goals in rare diseases research

200 New Therapies

Means to Diagnose Most Rare Diseases
How does it work?

- **Identify and define shared strategic goals**
- **Agree to approach jointly and pool resources**
- **Let each organisation use its own funding mechanisms/timing**
- **Agree to share data / standards**

Alignment – Flexibility - Commitment
Continued international cooperation

- International Human Metagenome Consortium
- International K.O. Mouse Consortium
- International Cancer Genomics Consortium
- International Rare Diseases Research Consortium
- International Human Epigenome Consortium
- Int. Initiative for Traumatic Brain Injury Research
IRDiRC timeline

- **Exploratory Workshop Reykjavik**
- **Launch Washington**
- **Working Groups**
- **Scientific Committees**
- **Support-IRDiRC Scientific Secretariat**
- **IRDiRC 2013 Conference**
- **2010**
- **2011**
- **2012**
- **2013**
- **2015**
- **2020**

- **3000 diagnostics**
- **6000 diagnostics**
- **50 new applications for market authorisation**
- **200 new applications for market authorisation**
IRDiRC Governance Structure

Executive Committee
Chair Ruxandra Draghia-Akli
Incoming: Paul Lasko

Scientific Committees
Diagnostics
Chair Kym Boycott
Interdisciplinary
Chair Hanns Lochmueller
Therapies
Chair Josep Torrent-Farnell

Working Groups

EU-funded SUPPORT-IRDiRC provides organisational support for Scientific Committees and Working Groups
Coordinator: Prof. Ségolène Aymé, INSERM
IRDiRC Governance Document

- Defines the aims of the consortium; mandate and composition of committees and working groups; rules for procedures, nomination of members, conflicts of interest etc.

IRDiRC Policies and Guidelines

- Will establish a common policy framework addressing various aspects of research, such as, sharing of data and samples, common quality standards, interoperability and harmonisation of ontologies and bio-banks, rapid release of research results, etc.
32 committed members

**Europe**
- E-RARE Consortium (EU)
- European Commission (EU)
- EURORDIS (EU)
- French Association against Myopathies (FR)
- French National Research Agency (FR)
- German Federal Ministry of Education and research (DE)
- Italian Higher Institute of Health Research (IT)
- Italian Telethon Foundation (IT)
- Lysogene (FR)
- Netherlands Organisation for Health Research and Development
- Prosensa (NL)
- Spanish Carlos III Health Institute (ES)
- UK National Institute for Health Research (UK)

**North America**
- Canadian Institutes for Health Research (CA)
- FDA Orphan Products Grants Program (US)
- Genome Canada (CA)
- Genetic Alliance (US)
- Mendelian Disorders Genome Centres (US)
- National Centre for Translational Sciences (US)
- National Cancer Institute (US)
- National Institute of Neurological Disorders and Stroke (US)
- National Institute of Arthritis and Musculoskeletal and Skin Diseases (US)
- National Institute of Child Health and Human Development (US)
- National Eye Institute (US)
- NKT Therapeutics (US)
- NORD (US)
- Office of Rare Diseases (US)
- PTC Therapeutics (US)
- Sanford Research (US)
- Shire (US)

**Asia**
- BGI (CN)

**Australia**
- Western Australian Department of Health (AU)
Diagnostics Scientific Committee

• Andrea Ballabio
• Michael Bamshad
• Kym Boycott (CHAIR)
• Han G. Brunner
• Xavier Estivill
• Milan Macek
• Gert Matthijs
• Frans Nauwelaers
• Peter Propping
• Jun Wang
Therapies Scientific Committee

- Gert-Jan B. Van Ommen
- Giles Campion
- Adam Heathfield
- Maria Mavris
- Fulvio Mavilio
- John McKew
- Elizabeth McNeil
- Luigi Naldini
- Glen H. Nuckolls
- Melissa Parisi
- Karin Rademaker
- Robert Schaub
- Joseph Torrenti Farnell (CHAIR)
- Mark Walton
- Ellen M. Welch
- Jack Welch
Interdisciplinary Scientific Committee

- Angel Carracedo
- Jamel Chelly
- Jack Goldblatt
- Alastair Kent
- Jeffrey Krischer

- Bartha Knoppers
- Hanns Lochmüller (CHAIR)
- Samantha Parker
- Rumen Stefanov
- Domenica Taruscio
Working groups to advise the Scientific Committees

- Ontologies
- Sequencing
- Model systems
- Genomics
- Bioinformatics
- Bio-banks
- Registries
- Biomarkers
- Ethics
- Repurposing drugs
- Therapies
- Regulatory issues
# Conference programme in a nutshell

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<th>Time</th>
<th>16 April</th>
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<tr>
<td>9:00</td>
<td>Plenary session</td>
<td>Therapies, Diagnostics &amp; Interdisciplinary tracks</td>
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<td>10:00</td>
<td>Networking Lunch</td>
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<td>11:00</td>
<td>Therapies, Diagnostics &amp; Interdisciplinary tracks</td>
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<td>Poster session &amp; dinner</td>
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Thank you & Welcome!

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http://ec.europa.eu/research/health