

IRDIRC NOVEMBER 2016 UPDATE



The program for the 3rd IRDiRC Conference is now available online

The 3rd International Rare Diseases Research Consortium (IRDIRC) Conference, will be held on February 8-9, 2016, in Paris, France. The conference gathers stakeholders from around the world to share their experience, knowledge and points of view to shape and advance rare diseases research through foundational, diagnostics, therapeutics and policy development. The full listing of speakers is available online on the [IRDIRC Conference website](#).

Confirmed speakers for the 3rd IRDiRC Conference include:

- Christopher Austin, National Center for Advancing Translational Sciences (NCATS), USA
- Claudio Bordinon, MolMed S.p.A., Italy
- Kym Boycott, Children's Hospital Eastern Ontario, Canada
- Ruxandra Draghia-Akli, European Commission – DG Research and Innovation (DG RTD), Belgium
- Jonathan Goldsmith, US Food and Drug Administration (FDA), USA
- Melissa Haendel, Monarch Initiative and Oregon Health & Science University, USA
- Edmund Jessop, National Health Service (NHS) England, UK
- Sangeeta Jethwa, Roche Innovation Centre, Switzerland
- Matt Might, University of Utah, USA
- Sophie Nicole, Université Pierre et Marie Curie, France
- Anders Olauson, Ågrenska Foundation, Sweden
- Slavé Petrovski, University of Melbourne, Australia
- Kiran Reddy, Clarus Ventures LLC, USA
- Makoto Suematsu, Japan Agency for Medical Research and Development (AMED), Japan
- Toshiki Takenouchi, Keio University, Japan
- Sonia Vallabh & Eric Minikel, Broad Institute, USA
- Nan Wu, Peking Union Medical College Hospital, China

The strong and exciting program, now [online](#), consists of 4 plenary and 3 parallel sessions. The conference kicks off with an overview of the history and achievements of IRDiRC five year after it was launched. This will be followed by a global view of rare diseases research in 2017, and the state of foundational, diagnostics and therapeutics research.

The first parallel session elaborates further the state of research today, with interesting talks on topics including data standards and exchange, undiagnosed diseases and ex-vivo stem cell therapy. The second

parallel session focuses on new approaches to rare diseases and highlights the work of innovative young investigators. The third parallel session provides an overview of trends in the fields of patient advocacy, companies, and regulatory and access.

The conference closes with a forward look plenary to discuss ways to transform rare diseases research over the course of the next 10 years, including a panel discussion on the next set of IRDiRC goals for 2017-2027. Participation and input of the community are vital so we can work together to bring diagnoses and therapies to all rare diseases patients.

Register [now](#) and join us in carving out the future of rare diseases research. Abstract submission and early registration deadline is extended to December 7, 2016.

We look forward to welcoming you in Paris, France, in February 2017!

Upcoming IRDiRC teleconferences and meetings

- December 7, 2016 – Operating Committee – Teleconference
- December 8-9, 2016 – Participant Unique Identifiers Task Force – Workshop, Paris, France
- February 6-7, 2017 – Consortium Assembly and Scientific Committees – Face-to-face meeting, Paris, France
- February 8-9, 2017 – 3rd IRDiRC Conference - Paris, France

Rare disease research published on the website

- International Recommendations for [undiagnosed patients](#)
- European Member States collaborate to [negotiate prices for orphan drugs](#)
- Challenges raised by [cross-border testing](#) of rare diseases in the European union

Research highlights from IRDiRC members

- [NGO Committee for Rare Diseases](#) launches at UN Headquarters
- YposKesi, the 1st French industrial pharmaceutical company for producing [gene and cell therapy drugs for rare diseases](#)
- 17th [rare disease walk](#) for the French Foundation for Rare Diseases
- Shire to establish [rare disease innovation hub](#) in Cambridge, Mass.
- From epigenetics - new molecular switches to [silence genes](#)
- [Undiagnosed Diseases Network](#) sticks around
- [Vasculitis](#): a rare-but-serious family of diseases you may never have heard of
- [Sanofi Genzyme](#) begins pivotal Phase 3 trial of NeoGAA investigational second-generation therapy for Pompe disease
- New technology to study human health advances goal of [regenerative medicine](#)
- 100 days to [Rare Disease Day 2017!](#)

Research highlights

Exciting workshop on Data-Mining and Repurposing

On November 16, a workshop of IRDiRC's Task Force on Data-Mining and Repurposing (DMR) took place in Barcelona, Spain, at the Barcelona Science Park. The purpose of this Task Force was to gather the expertise on data-mining and repurposing at global level and identify opportunities for collaborations, especially public/private ones, to speed up the exploitation of these new discovery tools. The workshop started with a discussion on the current state-of-the-art of the topics, quickly going over the major successes of data-mining and repurposing to date. The workshop continued with an evaluation on tools for assessing validity, followed by an analysis of *in silico* approaches versus experimental



approaches. Important debate was also held on sharing strategies and improved data sharing to help focus limited resources. The workshop ended with a summary of the workshop discussions and recommendations.

Dr Diego Ardigò elected as chair of the Therapies Scientific Committee

IRDiRC is pleased to announce that Dr Diego Ardigò has been elected the Chair of the Therapies Scientific Committee (TSC). Dr Ardigò is project leader of advanced therapy medicinal products (ATMP) and biologics at Chiesi Farmaceutici S.p.A. in Italy. Dr Ardigò will be replacing Mr Yann Le Cam, who is stepping down after serving a 3-year term and driving forth the establishment of several IRDiRC Task Forces. IRDiRC thanks Mr Le Cam for his commitment to IRDiRC, the TSC and patient voices, and we are pleased that he will continue to serve in the TSC.

IRDiRC-related calls

Fondazione Telethon funds a limited number of research projects in basic and clinical research ultimately aimed at [finding therapies for genetic diseases](#). This call is in support of research projects focusing on diseases of proven genetic origin, of either monogenic or polygenic forms.. Application deadline: January 18, 2017.

E-Rare has launched their call for [Transnational Research Projects for Innovative Therapeutic Approaches for Rare Diseases](#). The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear translational research approach. Deadline for pre-proposal submission: February 1, 2017.

Other news

If you are presenting in a meeting or a conference, and you would like to show some information about IRDiRC, standard slides are available on the IRDiRC private website; additional slides can be made available upon request. Please also email the Scientific Secretariat when and where you will be presenting, so we can keep track of "IRDiRC" presence at conferences.

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